

# The experience of individuals with Huntington's disease in the Western Cape, South Africa

By

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### **Declaration**

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## **Abstract**

**Aim:** The aim of this qualitative study was to explore the experiences of individuals with Huntington's disease (HD) in a South African context. The focus of the current study was not only on the challenges faced by individuals with HD, but also the resources/supports that help them cope with their neurological condition.

**Method:** I conducted twelve semi-structured interviews with the participants and they transcribed verbatim. I then performed a thematic analysis.

**Results:** Using Bronfenbrenner's Ecological System's Theory as the theoretical framework, several themes were identified that related to the participants' experiences of living with HD. Challenges included: triad of symptoms, sleep problems, testing process, relationships, children, it's a monster, employment, social support, partners and family members with HD, medical aid, life insurance, financial problems, lack of HD facilities, lack of understanding of HD, symptoms watching and the progression of HD. Several supports/resources were also identified and included: knowledge about HD, counselling, medication, coping, employment, social support, testing process, partners and family members with HD, medical aid, life insurance, cure, possible HD facilities, religion, grant and adaptation over time.

**Conclusion:** This was the first study of this kind in a South African context, which set out to explore the experiences of individuals with HD in the Western Cape, South Africa. The findings from this study demonstrate that although these individuals with HD experience several challenges due to their debilitating condition, they also employ several resources to help them cope with HD. Lastly, the findings that emerged from this study contribute to

raising awareness about the experiences of these individuals living with HD and could serve as a valuable foundation for tailor-made interventions for these unique individuals.

*Keywords: Huntington's disease, South Africa, challenges, resources*

## **Opsomming**

**Doel:** Die doel van hierdie kwalitatiewe studie was om die ervaringe van individue met Huntington se siekte (HS) in 'n Suid-Afrikaanse konteks te verken. Die fokus van die huidige studie het verband gehou nie net met die uitdagings wat deur individue met HS ervaar word, maar ook die hulpbronne / ondersteuning wat hulp verleen met hulle neurologiese toestand om dit te beter te hanteer.

**Metode:** Ek het twaalf semi- gestruktureerde onderhoude met die deelnemers gevoer en woordeliks getranskribeer , waarna tematiese analise uitgevoer is.

**Resultate:** Met behulp van Bronfenbrenner se Ekologiese Sisteem Teorie as die teoretiese raamwerk , is verskeie temas wat verband hou met die deelnemers se ervarings van die lewe met HD geïdentifiseer. Uitdagings sluit in: drietal van die simptome, slaap probleme, toets-proses, verhoudings, kinders, dit is 'n monster, indiensneming, sosiale ondersteuning, metgeselle en familie-lede met HD, mediese fonds , lewensversekering, finansiële probleme, die gebrek aan HD fasiliteite, 'n gebrek aan begrip van HD, dophou van simptome en die vordering van HD. Verskeie ondersteuning / hulpbronne is ook geïdentifiseer en sluit in: kennis oor HD, berading, medisyne, hantering, werk, sosiale ondersteuning, toetsproses, vennote en familie -lede met HD, mediese fonds, lewensversekering, genesing, moontlike HD fasiliteite , godsdiens, staats-toelaag en aanpassing oor tyd.

**Gevolgtrekking:** Dit was die eerste studie van hierdie aard in 'n Suid-Afrikaanse konteks wat die ervarings van individue met HD in die Wes-Kaap , Suid-Afrika uiteensit. Die bevindinge van hierdie studie toon dat, alhoewel hierdie individue met HD verskeie uitdagings as gevolg van hul aftakelende toestand ondervind, is daar ook 'n paar hulpbronne

in plek om hulle met die hantering van hierdie neurologiese toestand te help. Laastens , die bevindinge uit hierdie studie dra by tot die verhoging van bewustheid oor die ervarings van hierdie individue wat met HD lewe en kan as 'n waardevolle fondament vir pasgemaakte intervensies vir hierdie unieke individue dien.

*Keywords: Huntington se siekte, Suid-Afrika , uitdagings, hulpbronne*

## Glossary of terms

**Challenges:** Collectively, these are the various difficulties that individuals experience as a direct result from their HD.

**Chorea:** A neurological disorder, seen in Huntington's disease, characterised by involuntary, jerky movements seen in the shoulders, hips and face.

**Chromosome:** A thread-like structure transmitting the genetic information in most living cells, in the form of genes.

**Etiology:** The cause or set of causes of a condition or disease.

**Gene:** A unit of hereditary information transferred from parent to offspring that will determine several characteristics of the offspring.

**Huntingtin:** This is the gene, otherwise known as HTT or HD gene, which codes for a specific protein called huntingtin protein, produced in the brain. This protein is responsible for HD symptoms.

**Huntington's disease:** A hereditary, neurological disease marked by the progressive degeneration of brain cells, causing chorea and later, dementia.

**Participants:** Individuals recruited for this specific study who either had received a positive result on their predictive test or who have been diagnosed with HD and who resided in the Western Cape, South Africa.

**Predictive testing:** The predictive test was developed in 1993, and it is able to test an individual who is at risk of developing HD, by way of seeing if the individual carries the mutant gene, before they develop any symptoms of HD.

**Resources:** Collectively, those support structures that help individuals with HD cope with their condition.



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## **Chapter 1**

### **Introduction**

#### **1.1. Overview of Huntington's Disease**

Huntington's disease (HD) is a chronic, neurodegenerative disease that results in the loss of brain cells (Khalil et al., 2012). The loss of brain cells lead to a triad of symptoms, including motor disturbances, cognitive dysfunction and behavioural symptoms (Frank & Jankovic, 2010). The onset of the disease is usually between the 3rd and 5th decade of a person's life and affects females and males equally (Frank & Jankovic, 2010). Progression of HD is continuous with no periods of remission and apart from symptom relief, there is unfortunately no cure (Gudesblatt & Tarsy, 2011; Roos, 2010). HD is caused by a defective gene, called huntingtin gene (HTT) and has been linked to chromosome 4, which is inherited from either parent (Gudesblatt & Tarsy, 2011; Roos, 2010).

The incidence and prevalence for HD worldwide are estimated at 0.38 per 100 000 per year and 2.71 per 100 000 respectively (Pringsheim, Wiltshire, Dykerman, Steeves & Jette, 2012). The last prevalence study of HD in South Africa was in 1980, which found the prevalence to be 0.1 per 100 000 (Hayden, Macgregor, & Beighton, 1980). These statistics demonstrate that HD should be considered as a public health concern and is therefore seen as a research area worth investigating. Individuals with HD face several related challenges including: psychological reactions towards predictive testing, depression, gait and postural instability and impaired emotion recognition (Dalton et al., 2012; Gudesblatt & Tarsy, 2011; Ille et al., 2011; Reilman et al., 2012; Simpson, 2004; Smith, Mills, Epping, Westervelt, & Paulsen, 2012; Wetzel et al., 2011; Wiggins et al., 1992). On the other hand, there are several resources/supports that help these individuals to cope with their disease, such as a predictive

test support groups both in-person and online and physical exercise (Bohlen et al., 2013; Coulson, Buchaman, & Aubeeluck, 2007; Family Caregiver Alliance, 2003; van 't Spijker & ten Kroode, 1997).

The aim of this study is to explore the experiences of individuals with HD. More specifically, there will be a focus on both the challenges and the resources/supports experienced by these individuals in order to help them cope with their HD.

## **1.2. Overview of chapters**

Chapter 2 will look more closely at Huntington's disease, including the gene itself that causes HD as well as the signs and symptoms of this condition. Thereafter, chapter 3 will expand on the methodology that was used in this study, including the research design, participants, procedures, data collection and analysis, trustworthiness and ethical considerations. The results will be reported in Chapter 4, including the different themes and sub-themes that emerged from the data analysis. Lastly, Chapter 5 consists of the discussion of both the challenges faced and supports utilised by the participants. Both strengths and limitations of this study and suggestions for future research will also be discussed.

## **Chapter 2**

In this chapter, the relevant literature will be discussed. Firstly, the background of HD will be provided, where after the etiology as well as the clinical picture of HD will be reviewed. The clinical picture of HD consists of motor, cognitive and behavioural symptoms. Then the diagnosis, predictive testing and the treatment options will be covered, which will be followed by the stages of HD progression. After that, I will report on the literature examining the experiences of living with HD in terms of challenges and support structures. Lastly, the

theoretical framework, which is Bronfenbrenner's Ecological System Theory, will be described.

## **Literature review**

### **2.1. Background to Huntington's Disease (HD)**

HD is a progressive, neurodegenerative disease, which results in the slow loss of the affected brain nerve cells (Halpin, 2011). It is an inherited genetic condition that is clinically characterised by a triad of motor disturbances, cognitive dysfunction and behavioural symptoms (Frank & Jankovic, 2010; Halpin, 2011; Lundbeck, 2013). HD is autosomal dominant, meaning a child with a parent with HD has a 50% chance of inheriting the disease (Halpin, 2011; Ille et al., 2011; Williams, Skirton, Barnette & Paulsen, 2011).

Although Huntington's disease is classified as a rare disease, it occurs worldwide and crosses all racial and ethnic boundaries (Gudesblatt & Tarsy, 2011; World Health Organisation, 2013). However, there are differences between these rates among Western and Asian countries (Frank & Jankovic, 2010; Pringsheim et al., 2012). North America, Australia and Europe have a prevalence and incidence rate of 5.70 per 100 000 and 0.11 – 0.80 per 100 000 per year, respectively (Pringsheim et al., 2012). These statistics, are however, lower in Asia, as the respective rates are 0.40 per 100 00 and 0.046 – 0.16 per 100 000 per year (Pringsheim et al., 2012). These statistics confirm that it is a rare disease; however, if you include the close relatives and caretakers of individuals with HD then there are many more people who are affected by this disease (Quarrel, 2008; Williams et al. 2011).

If the onset of HD is before the age of 20 years, then it is called Juvenile HD and has generally been associated with paternal transmission (Gudesblatt & Tarsy, 2011; Haddad & Cummings, 1997; Kromberg et al., 1999).



Although HD is considered to be a rare disease (Williams et al., 2011), it not only affects the individuals with HD, but also the caregivers and other family members, both who are at risk and even those who are not. This makes the population who is affected by HD greater than only the sick individuals. Not only are some individuals overlooked where HD is concerned, but because HD is rare, it is sometimes also not researched in-depth. However, the intensity of the symptoms and the consequences of the disease create a need to investigate HD further, especially from a psychological standpoint.

## **2.2. Etiology**

George Huntington was the first to describe this eponymous disease in 1872, calling it “hereditary chorea” (Gudesblatt & Tarsy, 2011; Roos, 2010). However, chorea (abnormal motor movements) refers to only one of the triad (cognitive and behavioural symptoms) of manifestations of this disease, leading to a new term Huntington’s disease, which was thought to encompass all of the symptoms (Haddad & Cummings, 1997; Roos, 2010).

It was only in 1983 that the short arm of chromosome 4 was linked to HD and 10 years later in 1993, the specific gene for HD was identified (Gudesblatt & Tarsy, 2011; Roos, 2010). The Huntington’s Disease Collaborative Research Group suggested that the defective huntingtin gene (HTT) that was linked to HD contains CAG trinucleotide repeats (Gudesblatt & Tarsy, 2011; Huniche, 2011). CAG (cytosine (C), adenine (A), and guanine (G)), are three nucleotides that code for a specific amino acid, namely glutamine (Roos, 2010). Every person carries a number of CAG-repeats (up to 26); however, when there are 36 or more repeats, then they are associated with HD (Gudesblatt & Tarsy, 2011; Huniche, 2011). According to Gudesblatt and Tarsy (2011), if an individual has a gene in the intermediate region of between 27 and 35 repeats, then the gene may mutate into the range associated with HD in

his/her offspring. It has also been established that a correlation exists between the number of CAG-repeats and the age of onset of HD (Gudesblatt & Tarsy, 2011; Kromberg et al., 1999).

The discovery of the HD gene leads to the identification of the protein that it codes for, namely huntingtin (Quarrell, 2008.) Although the protein has been identified, the function of the protein is still not clear (Frank & Jankovic, 2010; Gudesblatt & Tarsy, 2011). It has been suggested that huntingtin play a role in intra-cellular transport and preventing neuronal toxicity (Frank & Jankovic, 2010; Gudesblatt & Tarsy, 2011). Individuals with HD, having the HTT gene, produce mutant huntingtin protein, resulting in abnormal cell physiology. This abnormal cell physiology presents itself as the clinical and pathological features of HD.

### **2.3. Clinical picture**

An individual with HD usually shows symptoms in three areas: problems with motor control, decline in cognitive functioning and psychological problems (Duff et al., 2010; Family Caregiver Alliance, 2003). These symptoms vary in number and intensity in every individual (Family Caregiver Alliance, 2003; Shoulson & Young, 2011).

#### **2.3.1. Motor symptoms**

Individuals with HD display a number of symptoms, one of them being movement problems. Chorea, abnormal involuntary movements, is the most distinct feature of HD and occurs in 90% of individuals with HD (Haddad & Cummings, 1997). Choreic movements include rapid, jerky movements, which the individual has no control over and affects distal extremities and facial muscles in the early stages of the disease (Gudesblatt & Tarsy, 2011; Ha & Fung, 2012). However, as HD progresses, signs of chorea usually becomes less apparent (Gudesblatt & Tarsy, 2011; Ha & Fung, 2012).

Ha and Fung (2012) reported that other motor symptoms in HD individuals include: dystonia (abnormal postures, twisting and repetitive movements as a result of sustained muscle contractions), postural imbalance, dysarthria (a motor speech disorder after injury to the speech area in the brain, resulting in weakened facial and mouth muscles, leading to poor articulation of phonemes) and dysphagia (having difficulty swallowing).

Another motor sign is their unique gait, which is interrupted by their choreic movements (Haddad & Cummings, 1997). Their manner of walking appears to be uncoordinated and individuals eventually find it impossible to walk (Haddad & Cummings, 1997).

Due to their motor symptoms, individuals with HD often have poor postural stability. As a result of this, they frequently experience falls (Haddad & Cummings, 1997; Ha & Fung, 2012; NetDoctor, 2012). Unfortunately, these recurrent falls are associated with higher morbidity and mortality rates, as well as fractures and head trauma (Haddad & Cummings, 1997; Ha & Fung, 2012).

### **2.3.2. Cognitive symptoms**

In addition to motor problems, individuals with HD also display several cognitive dysfunctions (Duff et al., 2010; Family Caregiver Alliance, 2003). Cognitive symptoms have been shown to be present at least 15 years prior to when a motor diagnosis is given and is directly related to the number of cell loss as a result of HD (Paulsen, 2011).

One of the cardinal signs of HD is dementia (Family Caregiver Alliance, 2003; Ha & Fung, 2012). Dementia is usually subcortical in nature and involves difficulties with executive functioning (involved in planning and organising), attention, language, memory, perception and visuospatial abilities (Paulsen, 2011).

Memory problems are frequently reported in individuals with HD (Bourne, Clayton, Murch, & Grant, 2006; Paulsen, 2011). There are three stages involved in memory, namely: taking in the information via senses, storing it and then recalling the information (Bourne et al., 2006). According to Ring and Serra-Mestres (2002), it is the third stage of memory – retrieving the information - that is problematic in individuals with HD. Difficulties with retrieving information applies to information stored a long time ago as well as to recently acquired information (Bourne et al., 2006). Implicit memory, collections of co-ordinated movement and skills that allow an individual to play a musical instrument, ride a bike or car, is also affected in individuals with HD (Paulsen, 2011).

Another cognitive sign or symptom that individuals with HD show is difficulties with executing executive functions, such as planning and organising of activities (Bourne et al., 2006; Haddad & Cummings, 1997; Lemiére, Decruyenaere, Evers-Kiebooms, Vandebussche & Dom, 2004). Individuals with HD perform poorly on tasks that test executive functioning, such as sorting cards by category and word fluency (Haddad & Cummings, 1997; Paulsen, 2011).

### **2.3.3. Behavioural symptoms**

Last of the triad of signs and symptoms are behavioural manifestations in individuals with HD (Family Caregiver Alliance, 2003; NetDoctor, 2012). Ha and Fung (2012) explain that behavioural symptoms of HD negatively impact the individuals quality of life, functional capacity, both leading to an increased risk of institutionalisation. The presence of behavioural or psychiatric signs or symptoms among individuals with HD varies from 35% to 73%, and includes a wide range of symptoms, such as: personality alterations, psychoses, mood disturbances and mixed disorders (Haddad & Cummings, 1997).

Cummings (1995) reported that alterations of personality are the most common behavioural symptom that individuals with HD present with. Some of these symptoms include irritability, emotional lability, apathy, aggressiveness and impulsiveness. In the study concerning individuals with HD conducted by Folstein (1989), 30.6% of the participants met the criteria for 'intermittent explosive disorder' and 5.9% were diagnosed with 'antisocial personality disorder'. Another study that looked at behavioural symptoms found irritability in 65%, apathy in 55%, and agitation in 59% of the participants with HD (Paulsen, Ready, Hamilton, Mega, & Cummings, 2001).

The second most common behavioural manifestation of HD is mood disorders, specifically depression (Family Caregiver Alliance, 2003; Haddad & Cummings, 1997). It has been reported that as many as half of individuals with HD suffer from depression (Pouladi et al., 2009; Rickards et al., 2011). Depression is most common in individuals with a later onset of HD and may precede motor symptoms (Haddad & Cummings, 1997). In addition to depression, anxiety, aggression and irritability, HD has been associated with suicidal ideation (Ha & Fung, 2012). The suicide rate among individuals with HD is 4 to 6 times higher than in the normal population, and the rate increases in individuals who are aged 50 or older (Haddad & Cummings, 1997).

#### **2.4. Diagnosis, predictive testing and treatment**

A diagnosis of HD is based on the clinical signs and symptoms in an individual who has a parent with proven HD (Roos, 2010). The first step for a diagnosis is to obtain a detailed history from the individual with symptoms, in addition to a detailed family history (Roos, 2010). When all the information has been gathered it is not difficult to make a diagnosis of HD, although symptoms that are non-specific can mislead the clinician (Roos, 2010). The clinical criteria that are currently used are motor symptoms, with or without

behavioural or cognitive signs or symptoms (Frank & Jankovic, 2010; Munoz-Sanjuan & Bates, 2011).

It is however possible to predict a diagnosis before the onset of symptoms, by means of a predictive DNA-test (PDT) (Coulson et al., 2007). A PDT was developed in 1993, capable of testing an individual who is at risk of developing HD, by way of seeing if the individual carries the mutant gene, before they develop any symptoms of HD (van 't Spijker & ten Kroode, 1997). Although a predictive test has positive functions such as giving the individual time to prepare for the future, only 15-20% individuals who are at risk of developing HD chooses to undertake the test (van 't Spijker & ten Kroode, 1997). Some of the reasons as to why these individuals choose not to be tested include: the fear of stigmatisation, discrimination in insurance and employment and the costs linked to testing as several genetic and psychological counselling sessions are required as well as specialised tests (Almqvist, Bloch, Brinkman, Craufurd, & Hayden, 1999; Bird, Bennett, & Lipe, 1993; van 't Spijker & ten Kroode, 1997). Other negative spinoffs of a PDT can include denying couples the opportunity to foster or adopt a child and individuals can lose their jobs in the armed forces as well as other professions (Simpson, 2004). A prenatal test, to see whether a foetus carries the HD gene, has also been developed with the aim to have the opportunity to terminate the pregnancy, although not many parents choose this option (Simpson, 2004).

Before the predictive test was developed, a probe (a short piece of DNA complimentary to a specific gene to locate it) called G8, was used as a predictive test but was not accurate enough (Connor, 1986). The very same probe that was developed in Boston in 1983 was first used in South Africa in the late 1980's (Magazi et al., 2008). In addition to the G8 probe used in South Africa, predictive tests have been offered in Johannesburg since 1989 and at the University of Cape Town from 1996 (Krause & Greenberg, 2008). Similar

programmes are now being offered in Durban as well as at Tygerberg Hospital in the Western Cape (Krause & Greenberg, 2008).

The predictive testing process for HD in South Africa has been designed in accordance to international testing protocols and includes a team that consists of a genetic counsellor, neurologist, genetic nursing sister, psychologist and psychiatrist (Krause & Greenberg, 2008). Pre-test consultations are the first step in this process and are used to establish the background history of the person who wants to be tested, to find out what their motivations are for undergoing the predictive testing and to inform them about HD (Kromberg et al., 1999). Blood is only drawn when the patient has been through all pre-test sessions (Kromberg et al., 1999). The results are given in person by a member of the predictive testing team, and at least one follow up session is provided if participants felt they needed more support (Krause & Greenberg, 2008; Kromberg et al., 1999). Sessions are also spaced out at 4-weekly intervals (Futter, Heckman & Greenberg, 2009), and include tests to evaluate depression and coping skills in individuals that want to be tested for HD (Kromberg et al., 1999). These guidelines, concerning who are involved, neurological and psychological tests and the timeline, have been developed with the aim of allowing individuals ample time to reflect on possible reactions towards their outcomes, making informed decisions regarding their motivations for testing and to reduce adverse emotional events (Futter, Heckman & Greenberg, 2009; Krause & Greenberg, 2008).

Unfortunately there is no cure for Huntington's disease yet (Gudesblatt & Tarsy, 2011; Roos, 2010). Therefore, the treatment of HD focuses on managing the symptoms with the use of a variety of drugs and therapies in order to improve quality of life for the individual with HD (Roos, 2010). In order to treat most of the motor symptoms, dopamine receptor blocking or depleting agents such as Tetrabenazine, which is approved by the Federal Drug

Association, are prescribed to individuals with HD (Frank & Jankovic, 2010; Roos, 2010). Individuals are also encouraged to make use of wheelchairs when going outdoors as falls can occur due to motor difficulties (Simpson, 2004). Concerning psychiatric symptoms, depression and aggression are the two main symptoms for which drugs are prescribed and include antidepressants and serotonin inhibitors (Haddad & Cummings, 1997; Roos, 2010). Other therapies that are recommended include: psychotherapy, speech-, physical and occupational therapy (Family Caregiver Alliance, 2003, Roos, 2010). Psychotherapy can provide guidance to help an individual with HD to manage their behavioural problems, facilitate effective communication between family members, and manage the individual's expectations of the progression of HD and to develop coping mechanisms (Mayo Clinic, 2011). Speech therapy is also useful to address issues with speech, swallowing and eating as it addresses the impaired muscles in the mouth and throat (Mayo Clinic, 2011). In addition to psychotherapy and speech therapy, physical therapy helps the individual with HD to increase their strength, balance, coordination and flexibility with appropriate exercises, which will maintain mobility for longer and may reduce the number of falls (Family Caregiver Alliance, 2003). Lastly, occupational therapy is recommended as it provides strategies to make homes more functional and safer for individuals with HD, such as installing handrails and assistance devices for bathing and dressing, and suggesting strategies to offset cognitive decline (Family Caregiver Alliance, 2003, Mayo Clinic, 2011).

## **2.5. Stages of HD progression**

The progression of HD can be roughly divided into three stages (a five-stage scale of progression is sometimes used by health care professionals) (Huntington's Disease Society of America (HDSA), 2013; Nance, Paulsen, Rosenblatt, & Wheelock, 2011). These stages were developed by Dr. Shoulson, a professor in Neurology, in 1979, in order for clinicians to



document the progression of the individual's HD (Paulsen, 1999). It must be noted that HD affects every individual differently, even members from the same family, and people will enter these stages described below at different times throughout the disease's progression (HDSA, 2013).

In the early stages of HD, individuals are mostly functional and may continue to drive, handle money, work, and live independently (HDSA, 2013). Symptoms that are present in this stage include minor involuntary movements, difficulty mentally working through complex problems, slight loss of coordination, and some may present with irritability, a lack of inhibition or depression (Nance et al., 2011).

Thereafter, in middle stage, individuals may lose the capacity to drive or work and may no longer be able to perform household chores or manage their own finances, but will be able to dress, eat and look after their personal hygiene with some assistance (Nance et al., 2011). Chorea may be prominent in this stage and individuals with HD may show increasing difficulty with performing voluntary motor tasks (HDSA, 2013; Nance et al., 2011). Additional problems may include balance, falls, swallowing, and weight loss (Nance et al., 2011). Individuals with HD in this stage also experience problem solving difficulties as they cannot organize, sequence, or prioritise information (Nance et al., 2011).

Lastly, individuals in late stage HD require assistance with all daily activities (Nance et al., 2011; Paulsen, 1999). Individuals in this stage are often bedridden and unable to form audible words, however, it is important to note that these individuals seem to retain some degree of comprehension (HDSA, 2013; Nance et al., 2011). Chorea may now be severe, but is most often replaced by rigidity, bradykinesia (abnormal slowness of movement) and dystonia (abnormal postures, twisting and repetitive movements as a result of sustained muscle contractions) (Nance et al., 2011). Psychiatric symptoms may also occur at any stage

during the progression of HD, but are unfortunately harder to recognize as well as treat in the later stages due to communication difficulties (Nance et al., 2011).

These stages were developed by Shoulson and Fahn (1979), based on the Total Functional Capacity (TFC), and are used to assess the functional status of the individual with HD, in order to reproduce a reliable and relevant indicator of the progression of the disease. The TFC rating scale has a range of 0 (minimal) to 13 (maximum) and assesses the individual's independence according to five domains: occupation, ability to manage their finances, capability to perform domestic tasks, capability to perform personal daily life activities and the level of care that must be provided (Nance et al., 2011).

## **2.6. Living with HD**

It is evident from the literature reviewed thus far that it must be challenging to live with HD. One of the aspects that this study will examine is the challenges that individuals who are diagnosed with HD face. Several challenges, faced by individuals with HD, have previously been identified and include: predictive testing, depression, poor cognitive performance, sleep disturbances, suicide, driving difficulties, decreased dependence, gait and postural difficulties, impaired emotion recognition and genetic discrimination. These challenges will all be discussed in more detail in the following sections.

One of the challenges that these individuals face is how to deal with the results of the predictive testing, especially if the outcome is positive (carrier of the HD gene) (Gudesblatt & Tarsy, 2011; Simpson, 2004; Wiggins et al., 1992). The emotional reactions towards a positive predictive test include: anger, denial, numbness, depression, anxiety and suicidal thoughts (Simpson, 2004; van 't Spijker & ten Kroode, 1997). A similar study conducted by Kromberg et al. (1999) in Johannesburg, South Africa that looked at predictive testing found

that 13% of the participants with a positive test experienced severe short-term depression. Other practical negative consequences of a positive outcome which are common are refusal of mortgages, insurance and critical life coverage (Gudesblatt & Tarsy, 2011; Simpson, 2004). Not only does a positive result have negative consequences, a negative result (not carrying the HD gene) can also have less desirable effects, such as the individual experiencing 'survivor guilt' (experiencing guilt as a result of not carrying the HD gene, while other family members have it) (van 't Spijker & ten Kroode, 1997).

Depression is not only related to the outcome of predictive testing, but is a common occurrence among individuals with HD (Smith et al., 2012). According to Smith et al. (2012) and Paulsen, Hoth, Nehl, and Stierman (2005), depression occurs in 40 to 50% of individuals with HD, with the prevalence decreasing with the progression of HD, although the severity of depression changes regularly. High rates of depression have been attributed to the fact that HD is a progressive, embarrassing, as well as disabling movement disorder, which occurs in the prime of an individual's life with serious cognitive deterioration and a 50% change of transmitting the gene to their offspring (Paulsen et al., 2005).

Poorer cognitive performance has been related to depression in previous studies where individuals with HD have participated (Nehl, Ready, Hamilton, & Paulsen, 2001; Paulsen et al., 2005, Smith et al., 2012). Smith et al. (2012) assessed cognitive performance, including processing speed and verbal performance skills by using several tests. Assessments for depression and cognitive performance included: the Beck Depression Inventory-II (BDI-II) to assess depressive symptoms, the Symbol Digit Modalities Test (SDMT) measures complex scanning, processing speed and working memory, the Trail Making Test A and B (TMT-A and TMT-B), where TMT-A measures psychomotor speed, speeded attention and visual attention, and TMT-B requires maintenance and set-shifting, the Stroop Color-Word

test to test the interference in reaction time of a certain task, and lastly, the Hopkins Verbal Learning Test-Revised (HVLT-R), both delayed (HVLT-DR) and immediate (HVLT-IR), which is a word list learning test (Smith et al., 2012). Participants with moderate and severe depression demonstrated poorer performance on all of the tests, except for the TMT-A and HVLT-DR tests, compared to individuals with minimal or mild depression (Smith et al., 2012). This suggests that depression could be associated with poorer cognitive performance in individuals with HD. Although the above mentioned studies include participants with HD who also experience depression, it should be noted that cognitive difficulties are one of the triad of symptoms in HD which can also occur independent of depression (Duff et al., 2010; Family Caregiver Alliance, 2003).

Another challenge that individuals with HD experience and that has been identified as a contributing factor to poorer cognitive performance, is sleep disturbances (Aziz, Anguelova, Marinus, Lammers, & Roos, 2010). One of the sleep disturbances which have been noted in individuals with HD is increased sleep onset latency (SOL). This is the time it takes for an individual to make the transition from full wakefulness to falling asleep. Other disturbances include frequently waking up during the night, reduced sleep efficiency and shortened and delayed rapid eye movement sleep (the lightest sleep stage where dreams occur). Lastly, it has also been noted that some individuals with HD experience increased periodic leg movements (a sleep disorder when an individual involuntarily moves limbs while asleep) either before falling asleep or during sleep (Arnulf et al., 2008; Aziz et al., 2010). Sleep disturbances, or lack of sleep, may also be considered to be a risk factor for the development of depression and is therefore considered a challenge (Aziz, et al., 2010, Videnovic, Leurgans, Fan, Jaglin, & Shannon, 2009). It was also reported in two studies, focusing on sleep problems in individuals with HD that these problems can potentially affect

participants' quality of life, may be a potential risk of injury, as well as may be a risk factor for depression (Arnulf et al., 2008; Aziz, et al., 2010).

Although sleep disturbances have been identified as a risk factor for depression, depression in itself has also been found to be a predictor of suicide, another challenge faced by individuals with HD (Wetzel et al., 2011). Suicide is an important clinical aspect of HD as the risk of suicide is increased in individuals with HD (Hubers et al, 2012). Successful suicide attempts has been recorded as high as 13% in individuals with HD, and there is a seven to twelve fold increase in the suicide rate among individuals with HD compared to the normal population (Wetzel et al., 2011). HD has the highest suicide rate compared to other neurodegenerative diseases (Wetzel et al., 2011). Factors contributing to this high suicide rate include: having to make the decision of wanting offspring with the risk of passing the gene on (Baliko, Csala, & Czopf, 2004), being unemployed (Almqvist et al., 1999), the presence of depression (Fiedorowicz, Mills, Ruggle, Langbehn, & Paulsen, 2011; Orth et al., 2010; Wetzel et al., 2011) and a history of psychiatric disorders (Almqvist et al., 1999). Other factors postulated by Hubers et al. (2011) are the emotional distress of having an incurable disease with a prolonged and devastating course, as well as experiencing diminished social support as affected family members pass away (Hubers et al., 2012).

Some studies have also examined predictors of suicide in individuals who carry the HD gene (Hubers et al., 2012; Wetzel et al., 2011). The study by Hubers et al. included 152 participants with the HD mutant gene, as well as 56 non-carriers. Suicidality was considered present if the individual had a score above 1 for the 'suicidal ideation' section in the Problem Behaviours Assessment (Hubers et al., 2012). The study found that having a formal DSM diagnosis of depression was a predictor of being suicidal. Also, predictive factors were studied by Wetzel et al. (2011) and it was found that that anxiety/depression and

irritability/aggression, in addition to substance abuse, were all predictive of suicide ideation. The study by Hubers et al. (2012) also supported previous findings of two critical periods of suicidality in individuals with HD, the first one being when at-risk individuals, who have not yet had a formal diagnosis, start to experience HD symptoms, while they include only non-specific neurological signs (Hubers et al., 2012; Paulsen et al., 2005). The second critical period of suicidality occurs when individuals with unequivocal HD signs become more dependent on others for their daily activities, as measured by the TFC (Hubers et al., 2012; Paulsen et al., 2005).

Another challenge experienced by individuals with HD is increased difficulty driving a car (Beglinger et al., 2012; Devos, Nieuwboer, Tant, De Weerd, & Vandenberghe, 2012; Rebok, Bylsma, Keyl, Brandt, & Folstein, 1995). Individuals with prodromal (before disease onset) and early stages of HD report a diminished ability to drive as the most frequent change in their day-to-day activities (Williams et al., 2011). The individuals with HD explained that they were concerned about their safety while driving, especially when they drove and got distracted or travelled at high speed (Beglinger et al., 2010; Williams et al., 2011). However, 70% of individuals continued to drive after the onset of HD (Rebok et al., 1995). Some reasons why people choose to stop driving is to prevent accidents, harm to themselves or even death (Devos et al., 2012; Haque, 2006). Automobile accidents can lead to increased financial burdens that the family must deal with, such as medical bills, insurance claims and loss of property (Haque, 2006). Another reason why individuals with HD cease driving is because of the way HD affects their mental capacity (Beglinger et al., 2012; Devos et al., 2012; Haque, 2006). Many individuals with HD experience problems with divided attention, which is the ability to split one's attention between more than one activity simultaneously (Haque, 2006). The result is that individuals with HD may not be able to concentrate on two

tasks at the same time whilst driving, consequentially meaning they might not stop at stop signs or red lights, they might be unable to remain in one driving lane at a time, and may engage in other accident-causing actions (Haque, 2006). The above mentioned actions can result from being distracted when driving, such as when having a conversation with fellow passengers, listening to music, or simply concentrating on something else than driving (Haque, 2006). In addition to safety and attention problems as reasons to stop driving, impaired implicit memory (a type of memory where past experiences help in the doing of a task without conscious awareness of these past experiences) in individuals with HD is another issue making driving more difficult (Haque, 2006; Paulsen, 2011). Implicit memories include coordinated skills and movements that allow someone to be able play a musical instrument, ride a bike and to be able to drive a car (Paulsen, 2011). Individuals with HD frequently have challenges with this kind of procedural, “unconscious” memory. As driving includes motor function, it fits into the implicit memory category and these individuals often find themselves getting lost and unable to follow direction (Haque, 2006). As a result, individuals with HD find that they require more conscious memory and concentration in order to drive a vehicle without problems (Haque, 2006). Lastly, movement difficulties also affect individuals with HD’s driving ability (Beglinger et al., 2012; Haque, 2006). Unfortunately, any uncoordinated or jerking movements of the driver, while he/she is driving, can lead to problems with stopping, turning, driving in a straight line or speeding (Haque, 2006).

As mentioned previously, individuals with HD have gait and postural difficulties, both posing challenges for them (Dalton et al., 2012; Reilman et al., 2012). As a result of postural instability and gait, loss of function and frequent falls has been observed in individuals with HD (Reilman et al., 2012). Reilman et al. (2012) noted that postural

impairment and motor difficulties increased as HD progressed. Dalton et al. (2012) examined how individuals with HD's gait and balance differed and found that symptomatic individuals presented with lower velocity, shorter stride and step length, less gait regularity and increased postural sway when compared to healthy individuals or pre-symptomatic individuals with HD. These difficulties make everyday tasks difficult and time consuming and are therefore the reason for functional loss (Dalton et al., 2012).

Individuals with HD also show impairment with regards to emotion recognition and experience (Ille et al., 2011; Novak et al., 2012). Ille et al. (2011) looked at emotion recognition in individuals with HD and found that, when presented with pictures showing facial expressions and having to rate the intensity of the emotion, individuals with HD rated intensity of anger, disgust and surprise lower than healthy individuals. Individuals with HD also struggle to correctly identify emotions shown in a picture (Ille et al., 2011; Novak et al., 2012). Moreover, pictures with affective scenes, which elicit happiness, fear or disgust, were rated more intense by individuals with HD than healthy control participants (Ille et al., 2011). These emotion recognition impairments can cause relationship breakdown as well as social isolation and is therefore regarded as a challenge faced by individuals living with HD (Novak et al., 2012).

Lastly, individuals with HD report that they experience genetic discrimination due to their condition (Bombard et al., 2009; Bombard et al., 2011; Erwin et al., 2010; Joly, Feze, and Simard, 2013; Pulst, 2009; Williams et al., 2010). Genetic discrimination can be defined as 'the denial of rights, privileges, opportunities, or adverse treatment based solely on genetic information, including family history or genetic test results' (Gostin, 1991, p. 12). For example, if an individual with a family history of HD want to take out life insurance, he/she might be denied or will have to pay an increased premium every month because of their risk



of developing HD. It has also been found that some people have been denied a job because of their family history of HD (Pulst, 2009). This definition includes a variety of discriminatory activities, such as being denied mortgagees, adoption rights, medical and life insurance that involve being treated differently after telling people about your HD status which individuals experience in their daily life as a result of a familial history or other discrimination and social stigma due to their genetic disease (Erwin et al., 2010). Genetic discrimination can be experienced both by individuals and their families and it is based on their actual or presumed genetic differences (Geller et al., 1996).

The studies by Bombard and colleagues (Bombard et al., 2009 Bombard et al., 2011) focus on genetic discrimination in Canada alone, whereas Erwin et al (2010) have investigated three developed countries, including Canada. Furthermore, Williams et al. (2010) builds on the Erwin et al. (2010) study and focuses on other aspects of discrimination, such as what individuals do in order to mitigate discrimination. For these reasons, the focus will be on the study by Erwin et al. (2010).

In the study conducted by Erwin et al. (2010) that investigated genetic discrimination among individuals with HD, it was found that a total of 46.2% of participants experienced some form of discrimination as a result of their HD. Genetic discrimination is deemed an important area of concern for not only patients and their family members who are at risk, but also for health care professionals and lawmakers (Erwin et al., 2010). Data was collected from Canada, Australia and the United States where 433 participants were recruited who were at risk for HD and either tested positive or negative for the mutant HD gene, and family members who have a 50% risk for developing HD but have not yet been tested (Erwin et al., 2010). Four categories of discrimination were evaluated and included: stigma or discrimination within relationships, employment discrimination, insurance discrimination,

and stigma or discrimination in transactions of daily living. The largest category of discrimination that was reported by participants (32.9%) occurred within personal relationships, which included changes in how people communicated with the participants, having negative comments to say to the participants, and discouraging participants to continue education. The category with the second most complaints was insurance as 25.9% of participants complained about this. Complaints mostly included being denied of insurance, such as life insurance, long-term care insurance, health insurance, disability insurance disability claim, auto insurance. In addition, 23.6% of participants stated how their genetic information was either accessed or asked for by insurance companies without their consent (Erwin et al., 2010). Erwin et al. (2010) noted that employment was the third most common category for genetic discrimination as a total of 26% of participants reported some form of discrimination while either at work or looking for a job. Experiences of discrimination included being denied a job, being fired, being covertly watched and being denied a promotion (Erwin et al., 2010). The last category of discrimination was concerning the participants' daily transactions with health care providers, the legal system, housing and other areas of daily life (Erwin et al., 2010). Although only a small amount of participants (4.6%) reported this as a challenge, it affected major life decisions, as some were denied the following: custody of their children after a divorce, adopting a child, donating blood, mortgages and/or renting a house (Erwin et al., 2010).

As mentioned above, individuals with HD face several challenges such as dealing with predictive testing, depression, poor cognitive performance, sleep disturbances, suicide, driving difficulties, decreased independence, gait and postural difficulties, impaired emotion recognition and genetic discrimination. However, these individuals also have access to several supports/ resources to help them cope with their disease. Some of these resources

include: predictive testing, assistive devices, support groups, both in-person and internet-based, gardening, occupational therapy and physical exercise

Although it was previously mentioned that predictive tests have negative spin-offs, van 't Spijker and ten Kroode (1997) found that predictive tests can also act as a form of support for individuals who suspect that they have HD. It was found that initial reactions towards a positive result include numbness, depression, anxiety and sadness, however, these feelings only lasted for a relatively short period of time and in most individuals returned to normal level within 12 months of the test (van 't Spijker & ten Kroode, 1997). One positive reaction toward a PDT is that individuals stated that their uncertainty about whether they are a carrier of the mutant gene was over and that would in turn result in relief (van 't Spijker & ten Kroode, 1997). Another positive reaction towards a PDT is that individuals who are at risk of HD said that they would enjoy their healthy period of life more after the test (van 't Spijker & ten Kroode, 1997). Lastly, it was also found that individuals who took the HD PDT said that the test would help them to establish what is really important in life once they knew what the outcome of the results are (van 't Spijker & ten Kroode, 1997).

As previously mentioned, individuals with HD experience difficulties with gait and balance, and as a result, frequent falls and injuries occur among these individuals (Bilney, Morris, Churchyard, Chiu, & Georgiou-Karistianis, 2005; Hausdorff, Cudkowicz, Firtion, Wei, & Goldberger, 1998; Kloos, , Kegelmeyer, White, & Kostyk, & Beyer, 2012). However, assistive devices, such as walkers and canes are often prescribed in order to prevent falls (Kloos et al., 2013). Kloos et al. (2012) systematically examined the effects of 6 different types of assistive devices (a cane, a cane with a weight, a standard walker, as well as a 2,3 and 4 wheeled walker) while measuring differences when walking in a straight line as well as around obstacles. There were 21 participants that performed both spatial and temporal tasks

and they were timed and observed for number of falls and stumbles while completing the course that was in a figure-eight pattern (Kloos et al., 2012). Kloos et al. (2012) concluded that walking was consistently better among participants with HD that used the four-wheeled walker compared to having no assistive device. Participants also had the lowest number of falls and stumbles and also walked the fastest when they were using the four-wheeled walker while doing the figure-of-eight course (Kloos et al., 2012). It should be noted that some of the other assistive devices made walking worse. This seems to suggest that although assistive devices can be a great source of support for people with HD, it is imperative that clinicians prescribe the appropriate assistive devices because these individuals have particular difficulties and needs (Kloos et al., 2012).

Another source of support for individuals with HD is being part of support groups (Family Caregiver Alliance, 2003). As a result of the strong emotional effect of a diagnosis such as HD, which is chronic and hereditary, and the constant stress that other family members may be at risk, involvement in a support group could be very helpful (Family Caregiver Alliance, 2003). Support groups offer safe and caring environments where information can be shared about the experience of living with HD and the accompanied challenges together with others in similar situations (Family Caregiver Alliance, 2003). Topics that are usually discussed include amongst other things, adjustment, disease course, coping, insurance, frustrations, family issues and medication (Family Caregiver Alliance, 2003). The Huntington's Association of South Africa (HASA) currently have support groups for individuals with HD in Johannesburg and Cape Town (Huntington's Association of South Africa, 2013). Monthly meetings are held and attended by individuals with HD, their caregivers as well as genetic counsellors and guest speakers who are experts on topics relevant to individuals with HD (Huntington's Association of South Africa, 2013).

A similar type of support or resource to support groups is the use of internet support groups (Coulson et al., 2007). Internet support groups are quickly becoming a frequently used resource, as a recent survey conducted by Pew Internet research institute showed that 36 million USA citizens had joined online support groups (Pew Internet research institute, 2005). An internet support group enables its users to partake in supportive interaction by means of bulletin boards, chat rooms, as well as individual email exchanges with others facing similar challenges or problems (Coulson et al., 2007). There are several advantages of such groups, the first one being that they are not restricted by geographical, spatial and temporal limitations that are typically experienced in face-to-face groups, meaning members can send and receive messages where ever they are, as well as at any time during the day (Pew Internet research institute, 2005). This is especially useful to individuals with HD as travelling for them is difficult as many of them stop driving because of cognitive and/or motor symptoms. Second, members can first carefully select the thoughts and ideas that they would like to share with the group at their own pace (Winzelberg et al., 2003). This is also ideal for individuals with HD, as previously stated, some cognitive functions in these individuals become slower as the disease progresses. The third benefit of online support groups is that they may bring together a wider range of individuals with more varied perspectives, opinions, experiences and sources of information that what would have been present in a face-to-face support meeting (Coulson, 2005). Fourth, there is a greater degree of anonymity offered by an online support group compared to face-to-face group meetings, which in return may facilitate self-disclosure and give members more confidence to discuss sensitive issues (Coulson & Knibb, 2007).

Coulson et al. (2007) examined the provision of social support stated in an online support group for individuals with HD, where 1313 messages were analysed through content

analyses. The authors concluded that both informational and emotional forms of social support were the most prevalent on the HD bulletin board (Coulson et al., 2007). The most frequent type of social support, as a fifth of the messages pertained to this form, offered by members of the online support group was informational support (Coulson et al., 2007). This included factual or technical information concerning HD, especially the genetic component of the disease, as well as offering advice on how to cope with all the different challenges posed by HD (Coulson et al., 2007). The second most prevalent form of social support was emotional support, as messages served an important part in acknowledging and validating others' views and experiences of HD. Emotional support was seen in messages that conveyed sympathy, empathy, encouragement as well as relief from blame to others (Coulson et al., 2007). HASA also provides this for their members as a website has been designed containing general information about HD, new articles regarding the development in HD and notices about upcoming support group meetings (Huntington's Association of South Africa, 2013). There is also a HASA Facebook page, where regular updates of recent fundraising events are broadcasted, as well as giving members the opportunity to post questions and answers on the page (Huntington's Association of South Africa, 2013). As typical of other support groups, HASA also plays an important role in emotionally supporting one another in the group, especially people who have been recently diagnosed or received their predictive test results (HASA, 2013).

A different form of resource that has been studied among individuals with HD is gardening as an activity for people with advanced HD (Cooper Marcus, 2007; Spring et al., 2011; Spring, Viera, Bowen, & Marsh, 2013). Gardening is an example of a non-pharmacological but practical form of intervention for people with HD (Spring et al., 2011). The publication by Spring et al. (2011) suggests that residents benefitted from the wide range

of movement-based and dexterity tasks, which required concentration and precision (Spring et al., 2011). Gardening introduced group cohesiveness as many residents worked together in the garden, which stimulated socializing amongst the residents (Spring et al., 2011). Spring et al. (2013) further expanded on the therapeutic benefits of gardening at a Huntington's centre in London, England, where they interviewed residents, staff as well as visitors. An occupational therapist assessed the activity to be safe for the individuals with HD to participate in, and a horticulturist adapted the gardening by raising the plant beds and providing small, light tools that were placed within easy reach (Spring et al., 2013). Benefits were reported by means of interviews and questionnaires from visitors and staff and a special pictorial questionnaire for individuals with HD (Spring et al., 2013). The residents with HD from the centre seemed to benefit from the garden and some of these benefits included: being happy and enjoying being in the garden, looking at birds, looking at the flowers, enjoying the process of growing plants, painting flower related pictures, and lastly watching garden related television programmes (Spring et al., 2013). Spring et al. (2013) also noted specific clinical cognitive and physical benefits. Cognitive benefits were giving the residents a sense of ownership, in addition to the opportunity to engage in sequencing (matching flowers by colour and shape) and problem solving. Physical benefits from gardening included having the opportunity for increased functional movement, good physical work, especially hand movement, and habituated patterns of movement (Spring et al., 2013). Spring et al. (2013) concluded that the inexpensive programme of activities enabled creativity and self-expression, stimulated social contact and aided the clients' therapeutic goals. The therapeutic opportunities provided by gardening include both movement and cognitive benefits and is thus a noteworthy resource for individuals with HD (Spring et al., 2011; Spring et al., 2013).

Occupational therapy (OT) has also been identified as an important resource for individuals with HD to cope with their disease, because occupational therapists aim to retain or reinstate that is beneficial for the individual in terms of his or her abilities, the demands of his or her occupation and the demands of their environment (Bilney, Morris & Perry, 2003; Steultjens, Dekker, Bouter, Leemrijse & van den Ende, 2004). Clients who are referred for occupational therapy can be of all ages, with mental, physical and/or social impairments, and/or learning challenges (Steultjens et al., 2004). Mason, Andrews, and Wilson (1991) described the beneficial effects of an OT program that was aimed at helping individuals carry out personal daily life activities, such as eating and washing. The intervention included re-educating each individual concerning personal care activities such as brushing teeth, drinking from a cup and washing ones face (Mason et al., 1991). The intervention was run over a course of 16-weeks, where after effectiveness was rated on a 9-point scale that rates the level of external assistance that is required by an individual in order to complete an activity (Mason et al., 1991). OT prevented any significant further deterioration in functional tasks performed by the individuals during the course of the intervention (Mason et al., 1991). Di Scipio and Hannesson (1971) studied the effects of OT provided to a single female participant with HD, which had to be nursed on the floor due to her inability to maintain a sitting position. After 10-weeks of 20 minute OT sessions, the participant could maintain a sitting position, as well as lift her head and shoulders from a prone position, as well as to ambulate (Di Scipio & Hannesson, 1971). Although the validity of this study is limited because it was a single-case study, OT could be helpful for individuals with HD (Di Scipio & Hannesson, 1971).

Lastly, physical exercise is also considered important to cope with HD as these individuals have problems with movement (Bohlen et al., 2013; Khalil et al., 2012). Such



strategies can be effective in allowing people with HD to maintain independence for longer, reduce the number of falls, which will result in improvements in their quality of life (Busse & Rosser, 2007). Bohlen et al. (2013) examined whether physical treatment could help with posture and gait problems in individuals with HD. The study included twelve participants who had to partake in a six-week predefined physical intervention, which included a warm-up session, different ways of walking, postural stability exercises, balance and gait training, motor coordination as well as relaxation exercises (Bohlen et al., 2013). Bohlen et al. (2013) found that participants needed less double support (both feet touching the ground), walked faster as well as having longer stride length than at baseline. This is significant as severe gait difficulties are the most important factor that nursing homes consider when deciding not to take in an individual with HD (Bohlen et al., 2013).

Zinzi et al. (2007) incorporated physical exercise into their intense rehabilitation programme for individuals with HD and also found that motor performance in these individuals increased. Forty participants were recruited who were either in the early and middle stages of HD (Zinzi et al., 2007). The physical therapy component included exercises in order to improve gait, balance, strength, coordination, posture and stability, which were administered in a three-week course, three times a year for two years (Zinzi et al., 2007). Both the Tinetti Scale and Physical Performance Test were used to assess motor performance after each 3-week admission (Zinzi et al., 2007). The Tinetti Scale is a 28-point scale that measures sitting and standing balance and the smoothness of gait while doing this, while the Physical Performance Test measures execution of functional tasks, such as picking up and storing object, and is timed to measure accuracy and speed (Zinzi et al., 2007). After each 3-week admission and completing the rehabilitation programme after 2 years, it was established

by Zinzi et al. (2007) that scores on both tests increased significantly, showing that physical exercise improves motor performance in individuals with HD.

Although physical exercise has been linked to increased motor performance, there are factors, including cognitive and motivational issues that may influence individuals with HD and their participation in exercise outside clinical settings where supervision is not provided (Khalil et al., 2012). Unfortunately, exercise adherence decreases after professional supervision stops and individuals no longer receive external support (Khalil et al., 2012). Therefore, home-based DVDs and video-games have been used to encourage individuals with HD to continue their exercise regime (Khalil et al., 2012; Kloos, Fritz, Kostyk, Young & Kegelmeyer, 2013;). The technology capabilities of these formats, such as subtitles, music, and visual and verbal cues, can be useful to facilitate engagement in exercise and to ensure correct execution of exercise (Khalil et al., 2012). More specifically, Kloos and colleagues (2013) administered a video game-based exercise, called Dance Dance Revolution, to a group of individuals with HD in order to see whether motor performance can be improved. Dance Dance Revolution requires players to step on appropriate arrows as they respond to visual cues, while matching a song rhythm (Kloos et al., 2013). This specific game incorporates attention strategies, balance training methods and external cueing (stepping in a rhythmic manner), which are all recommended for individuals with motor movement deficits (McIntosh, Brown, Rice & Thaut, 1997; Thaut, Miltner, Lange, Hurt & Hoemberg, 1999). Participants were evaluated after playing the game twice a week for six weeks and were found to spend significantly less time in double support position (both feet on the ground) when walking forward and backward (Kloos et al., 2013). Participants who had less severe HD motor symptoms also displayed reduced heel-to-heel base support (Kloos et al., 2013). These findings suggest that Dance Dance Revolution leads to improved dynamic balance

while walking and suggest that this type of interventions can be helpful to make life easier for people with HD (Kloos et al., 2013).

It is evident from the literature review that individuals with HD face several challenges as a result of their disease, and include: psychological reactions towards predictive testing, depression, poor cognitive performance, sleep disturbances, suicide, driving difficulties, gait and postural instability, impaired emotion recognition and genetic discrimination (Aziz et al., 2010; Berglinger et al., 2012; Dalton et al., 2013; Erwin et al., 2010; Gudesblatt & Tarsy, 2011; Ille et al., 2012; Nehl et al., 2001; Reilman et al., 2012; Simpson, 2004; Smith et al., 2012; Wetzel et al., 2011; Wiggins et al., 1992). However, there are several support/resources that help individuals with HD cope, for example a predictive test, support groups both in-person and online, assistive devices, gardening, occupational therapy and physical exercise (Bilney et al., 2003; Bohlen et al., 2013; Coulson et al., 2007; Family Caregiver Alliance, 2003; Kloos et al., 2012; Spring et al., 2013; van 't Spijker & ten Kroode, 1997). It is evident from the literature that there are some studies internationally that have explored the experiences of what it is like to live with HD. However, to my knowledge, no studies examining the experiences of individuals with HD have been conducted in South Africa. South African studies concerning HD until the present day have focussed on the epidemiological and genetic component of HD (Baine et al., 2013; Futter, Heckman, & Greenberg, 2009; Hayden & Beighton, 1977; Krause & Greenberg, 2008; Kromberg et al., 1999; Magazi et al., 2008; Silber, Kromberg, Temlett, Krause & Saffer, 1998).

## **2.7. Bronfenbrenner's Ecological System's Theory**

This study will be conceptualised within the context of Bronfenbrenner's Ecological System's Theory, or Human Ecology Theory (Bronfenbrenner, 1979). This theoretical framework will be used to explain the experiences of individuals living with HD, focusing on

the challenges and supports they encounter as a result of their condition. This theory describes human development in terms of five systems: micro-, meso-, exo-, macro- and chrono-system (Bronfenbrenner, 1979).

This framework has been chosen because the experiences of the participants with HD in this study will be looked at both at an individual, for example friends and families, as well as a contextual level, such as the different policies of medical aid schemes and life insurance companies relevant to them.

The first level in this theory is the micro-system, and is the person's immediate environment where he/she finds him/herself, where intimate and direct interactions occur, such as with friends, family, neighbours and work colleagues (Bronfenbrenner, 1979). Examples of these interactions on this level, which are relevant to this study, would be the relations between the individual with HD and his/her family, friends, work colleagues and medical staff during the testing process. Thereafter is the meso-system, which refers to the interactions or links between the micro-systems, where the individual actively participates (Bronfenbrenner, 1979). According to Bronfenbrenner, this would be, for example, the interactions taking place between the individual with HD's families and their partners/spouses. The next level is the exo-system, which includes those structures (from both micro- and meso-systems) that the individual is not directly implicated in, for example, the individual with HD's spouse's/partner's working environment (Bronfenbrenner, 1979). The macro-level follows, which encompasses culture, values, laws and policy making (Bronfenbrenner, 1979). Examples of this level would be medical aid schemes, life insurance and religion. The last level is the chrono-system, which includes the shifts and transitions in one's lifespan. One example of this would be how a divorce, as a major life transition, may affect the individual with HD's life (Bronfenbrenner, 1979).

From the current literature, it is evident that individuals with HD experience both challenges because of their condition, but also several supports/resources in order to help them cope. In order to aid in the discussion of challenges and supports/resources pertaining to individuals with HD, Bronfenbrenner's Ecological System's Theory will be used. Hereafter follows chapter 3 describing the current study's methodology.

## **Chapter 3**

### **Methodology**

This chapter will discuss the study protocol in more depth, which will include: the research rationale, the research question, aims and objectives, research design, participants, data collection, analysis, trustworthiness and the ethical considerations which were undertaken before the study commenced.

#### **3.1. Research rationale for this study**

It is important to conduct this research because HD is a debilitating lifelong disease, which includes physical and psychological symptoms (Roos, 2010). As a result of their disease, these individuals face many challenges and therefore it is essential to explore the experiences of individuals who are living with HD (Roos, 2010). When compared to other chronic diseases, the prevalence of HD is relatively low (Quarrel, 2008; Williams et al. 2011). This might be one of the reasons for the paucity of research that relates to HD. It should be noted that although the prevalence rate of HD is relatively low, it is not only the individual with HD that is affected, but also his/her family members that are at risk, as well as the caregivers who take care of individuals who suffer from HD. Although a few studies investigated aspects of the experiences of individuals living with HD (Family Caregiver Alliance, 2003; Gudesblatt & Tarsy, 2011; Reilman et al., 2012; van 't Spijker & ten Kroode, 1997), to my knowledge no studies of this nature have been conducted in South Africa. It can be expected that all these individuals with HD across the world will present with similar signs and symptoms, as it is mainly a medical condition that will be studied. However, contextual and/or cultural factors will affect the experiences of individuals with HD to a certain degree. The quality of support and services offered by health care services, as well as support

provided by family, friends and the community may vary from one context to another. For example, the study will set out to investigate the predictive process offered by Groote Schuur hospital, in the Western Cape. Other factors that may differ across contexts may include the availability of social support structures, such as the existence of support groups, and the accessibility and competence of medical care services, for example medical aid schemes and specialised caring facilities. Support from the South African government, such as grants and housing subsidies, may also differ greatly from other countries, such as the United Kingdom or Australia. Thus, the purpose of this proposed study is to investigate the experiences of individuals with HD, within the South African context, with a specific focus on the challenges and the factors that help them cope with this condition. Knowledge from this study can inform the planning and implementation of interventions for individuals who suffer from HD.

### **3.2. Research question**

This study will aim to address the following question:

- What are the experiences of individuals living with Huntington's disease in the Western Cape, South Africa?

### **3.3. Aims and objectives**

The purpose of this study will be to explore the experiences of individuals living with Huntington's disease, focusing on:

- The challenges that individuals with Huntington's disease experience; and
- The resources and/or supports perceived by the individuals as helpful to cope with Huntington's disease.

### **3.4. Research design**

This study was a qualitative, exploratory study. Bless, Higson-Smith and Kagee (2006) explain that exploratory research is appropriate when there is little knowledge available regarding a specific research topic or question and new insights into phenomena are needed. A qualitative approach is also applicable as this type of data will generate new and unforeseen information regarding this topic (Bless, Higson-Smith, & Kagee, 2006).

### **3.5. Participants**

I made use of purposive sampling. This type of sampling is appropriate to employ if the desired group of people, individuals with Huntington's disease (HD) in this case, is rare or difficult to recruit and locate (Bless et al., 2006). My participants for this study were individuals who have been diagnosed with HD. My supervisor, Dr. Pretorius, and I were invited to join a support group meeting of the Huntington's Association of South Africa (HASA) in Cape Town in February (see attached letter of invitation, (appendix 1). I presented the proposed research to the group, where after individuals were invited to take part in the study. Thereafter, a blank list was passed around so that individuals who wanted to participate could provide their contact details. The group members were informed that 60-90 minutes of their time would be needed for the interview.

To participate in the study, the diagnosis of HD must have been confirmed at least six months ago. I aimed to conduct semi-structured interviews with at least 15 individuals who live with HD. The precise number of interviews depended on when theoretical saturation was reached, i.e. when the same themes started to reoccur (Guest, Bunce & Johnson, 2006). However, according to Guest et al. (2006), theoretical saturation of themes during qualitative



analysis usually occurs within the first twelve interviews when the sample is homogenous, and could occur with as few as six interviews at times.

The withdrawal rate for study either before or during the interviews was 0%. The participant sample consisted of eight females (67%) and four males (33%), with ages ranging from 22 to 63 years (mean=41) (refer to Table 1). The time since diagnosis ranged between 1 year and 25 years (mean = 10 years).

Although HD may be associated with disability, the individuals who participated in this study ranged in their functional capabilities. Three of the participants could not work anymore due to severe HD cognitive symptoms, such as memory and concentration problems, whereas others still worked full-time and required little additional help from their spouses with daily activities.

Table 1

*Demographic details of participants*

Participant code	Age (yrs)	Race	Language	Years since diagnosis	Support group
M1	32	white	English	3	yes
F2	28	white	English	2	no
F3	30	white	English	2	yes
F4	40	coloured	English	9	yes
F5	34	white	English	4	no
F6	55	white	English	25	yes
F7	63	white	English	20	yes

F8	46	white	English	10	yes
M9	50	white	English	23	no
F10	31	coloured	Afrikaans	5	no
M11	58	coloured	Afrikaans	14	no
M12	22	white	Afrikaans	1	no

*Note.* Participant code: F=Female; M=Male;

### 3.6. Data collection

The potential participants who showed interest in taking part in this study were contacted either by e-mail or telephone, and were reminded about the nature of the study. The participants were again given the opportunity to choose whether they still wanted to participate. An appointment was arranged with all the participants at a location and time that suited them. Before the interviews started, informed consent from each participant was obtained (see Appendices 2 and 3). In addition to the informed consent, a short biographical questionnaire was also handed out to the participants to complete (see Appendices 4 and 5). Thereafter, the semi-structured interview followed, which consisted of broad open-ended questions pertaining to the experiences of individuals living with HD (see Appendices 6 and 7). I anticipated that the interviews would take between 60 and 90 minutes to conduct. All of the interviews were tape-recorded, with the participants' permission, in order to transcribe them verbatim to perform the necessary analyses.

### 3.7. Analysis

Thematic analysis was used to analyse the transcribed data. The first step of this method was to acquaint oneself with the collected data, by means of transcribing and reading the data (Braun & Clarke, 2006). For this reason, I transcribed all of the interviews myself.

After transcription was completed, each participant's transcribed interview was sent to him/her. This gave the participants an opportunity to either add or take away from their responses and confirm whether it was an accurate account of the interview. After that, initial codes, for interesting features of the complete transcribed data set, were generated (Braun & Clarke, 2006). I did this when I read through the transcripts, highlighting appropriate statements and adding initial codes in the margin. Thereafter, it was necessary to organise the possible codes into several themes, as well as to review the themes by examining whether they correlate against the complete set of data (Braun & Clarke, 2006). This was done by copying the highlighted extracts and pasting it into a new Word document where I grouped them into possible themes. Next, the themes were defined and named in order to refine each of the themes (Braun & Clarke, 2006). In addition to my identification of themes, my supervisor also did the previous steps independently to see whether the same themes emerged. Lastly, a report was produced, which included vivid, compelling extract examples of the interviews with the participants (Braun & Clarke, 2006). I used direct quotations from the participants' interviews to illustrate the identified categories and themes.

### **3.8. Trustworthiness**

Several measures were also taken to strengthen the trustworthiness of the current study. Lincoln and Guba (1985) posit that qualitative researchers should consider the following four criteria in order to pursue a trustworthy study: (a) credibility; (b) transferability; (c) dependability; and (d) confirmability. The first criteria, which is credibility, refers to accurate interpretations and descriptions of the participants' experiences and views by the researcher(s) and is argued to be the most important criteria ensuring trustworthiness (Lincoln & Guba, 1985). One strategy to improve a study's credibility is that the researcher(s) must be familiarised with the culture of the participating organisations

before data collection commences (Lincoln & Guba, 1985). This was done by first visiting several HASA meetings as well as building a relationship with the group leader. Not only was it necessary to familiarise myself with the participants but also previous literature regarding the topic (Lincoln & Guba, 1985). As part of this study, a full literature review was compiled before interviews with participants commenced. Another strategy to enhance credibility is triangulation, which is when a research team, opposed to a single researcher, is employed. In addition to myself, two other members of the research team independently performed all the steps of the data analysis process previously described. The themes that emerged from thematic analysis were compared and differences in interpretations were discussed and resolved through discussion of the themes. Consensus was reached by all three researchers and is reflected in the final version of the findings. Another way that credibility can be enhanced is using tactics to help ensure that participants are honest when contributing data to the study (Lincoln & Guba, 1985). This was achieved by firstly, reiterating that participation is voluntary and that they may refuse to proceed with the interview with no negative consequences, as well as explaining that everything will be anonymous, meaning no sensitive information could be traced back to them. Lastly, Lincoln and Guba (1985) state that credibility can be improved by having peers scrutinise the research proposal before collecting the data. As a standard process in the psychology department, at least two colleagues, usually with expertise knowledge about the particular field, are required to give feedback on the research proposal before the study is initiated.

The second criterion, transferability, refers to extent to which the findings of a particular study can be generalised to other situations (Lincoln & Guba, 1985). Sufficient contextual information about all the fieldwork should be provided by the researcher. Therefore, thick descriptions concerning the participants, such as inclusion and exclusion

criteria and how many participants were involved, as well as the research context and setting, were all described in order for the reader to assess the transferability of the findings. Lincoln and Guba (1985) maintain that it is the researcher's responsibility to provide a comprehensive database in order to make conclusions about the transferability about the study by others; however, it is not the researcher's obligation to provide an index of transferability within qualitative research.

Thirdly, dependability addresses the issue of whether the findings would be similar if the study was to be repeated by a different researcher (Lincoln & Guba, 1985). Lincoln & Guba (1985) state that by describing the research design and its implementation, explaining the data gathering process on an operational level and evaluating the effectiveness of the study are all ways to enhance dependability. All of these processes have been fully described and terms have been defined. The evaluation of the study will follow later, which will mention both strengths and weaknesses.

Lastly, trustworthiness includes confirmability, which is the extent to which the results of a study were shaped by the participants instead of researcher bias, interest, or motivation (Lincoln & Guba, 1985). Both triangulation and reflexivity were employed during this study to enhance conformability. Triangulation, as previously mentioned, is using more than one researcher to derive themes (Lincoln & Guba, 1985), and was used in this study. In addition to triangulation, reflexivity was also made use of in order to improve confirmability. Reflexivity refers to attitude of attending systematically to the effects of the researcher on the context of knowledge construction (Lincoln & Guba, 1985). Therefore, I kept a reflective diary as the primary researcher, and the content was discussed with two additional team members, one who is an experienced Huntington's disease researcher and the other who had

expertise knowledge concerning qualitative research. Thus, reflexivity was increased, as personal biases, perception and attitudes were discussed.

### **3.9. Ethical considerations**

Ethical approval, for the commencement of this study, was applied for at the Health Research Ethics Committee at the Faculty of Health Sciences at Tygerberg Hospital, and was granted with ethics reference number: S13/04/087 (Appendix 8). The invited participants were informed about the relevance and nature of the research. They were reminded that their participation was entirely voluntary as well as that they had the right to withdraw from the proposed study during any stage without experiencing any negative consequences. The participants were informed that they would incur no additional expenses if they wished to participate in the study. Informed consent, which included their permission to record the interview, was first required from the participants before the interviews started. Each participant was assigned a number when the data was analysed in order to protect the participants' anonymity. The participants' personal details were only shared between my supervisor and I. Once the tape-recorded interviews were transcribed, they were kept safe and secure in a locked drawer in Dr. Pretorius's office. The tape-recordings were destroyed and discarded in an appropriate manner when data-analysis was completed. The only risk that we anticipated was that the participants may experience emotional discomfort and/or distress during the interview due to the personal nature of the interview. If this was to be the case, I would then have referred the participants to my supervisor, Dr. Pretorius, who is a Counselling Psychologist. She would then have referred them to appropriate health care professionals. However, this was not necessary, as no one presented with emotional distress or expressed a need to be referred to a health care professional for additional services.

### 3.11. Self-reflection

I was the primary investigator during this study, under the supervision of Dr. Chrisma Pretorius. I am a 24-year old female Psychology Master's student. Having had a passion for psychology and the benefits of this profession, I enrolled in a Bachelors of Science (B.Sc) in 2009. My degree enabled me to combine psychology, which was one of my main subjects, with other scientific, biological subjects, such as genetics and physiology. I specifically chose this route as I thought that psychology and physiology would be in interesting combination, as sometimes psychological challenges can be a cause of physiological symptoms and vice versa.

After my B.Sc, I enrolled in the Psychology Honours programme at the University of Stellenbosch. This is where I first developed my interest for research and understood the importance of it. For my honours dissertation, I looked at Multiple Sclerosis and certain psychological aspects of the disease. I thoroughly enjoyed the process of recruiting participants, interviewing them, transcribing and analysing the data in order to compile a report. This research was greatly appreciated by the participants and the Multiple Sclerosis Society of South Africa, and as a result, I had the privilege of presenting my findings at several support groups at Medi-Clinic hospitals in the Western Cape as well as at their annual function.

However, after I completed this project I was eager to further my research career. I decided that I want to do my masters degree on HD, as I had previously witnessed what research means to a group of people with a serious medical condition who have received little attention in the past.

As I discovered, reflection is a fundamental aspect of qualitative research (Long & Johnson, 2000). Reflexivity requires the researcher to reflect on their own belief system, as they observe the beliefs of his/her participants (Long & Johnson, 2000). It is very possible that my age of 22 years could have impacted the interviews with my participants, as all of the participants were older. This may have caused the participants to withdraw from sharing sensitive information, such as sexual difficulties due to HD. However, I felt confident in conducting the interviews as I have had previous experience with qualitative interviews, having done qualitative interviews for my honours project as well as having received training during the honours course from qualitative research experts. However, although I researched HD extensively and had some neuropsychological background before commencing with the interviews, I still did not know everything about HD. Yet I do not believe that my age or lack of expertise negatively affected the results of this study as participants described the interviewing process as a positive experience.

## **Chapter 4**

### **Results**

Chapter 4 will display the results, which resulted from thematic analysis of the interview transcripts. All of the main and sub-themes follow below, with quotations in order to vividly describe them.

#### **4.1. Introduction**

From the semi-structured interviews conducted with the participants of this study, it emerged that despite differences in time since diagnosis and the severity of the condition, there were several themes that appeared to be common across the experiences of participants. The main themes and their respective sub-themes that emerged through the process of



thematic analysis are presented in Table 2. The two main themes consisted of the challenges faced by individuals living with HD and the resources which help HD individuals to cope with this progressive, neurological condition. The main themes, as well as related sub-themes which emerged through thematic analysis were conceptualised and structured according to Bronfenbrenner's ecological model (Bronfenbrenner, 1979).

Table 2

*Major and related sub-themes according to Bronfenbrenner's ecological model*

Main theme	System level of Bronfenbrenner's ecological model			
	Micro-system	Meso-system	Macro-system	Chrono-system
Challenges	Triad of symptoms	Partners and family	Medical aid	Symptom watching
	Sleep problems		Insurance policies	Progression of HD
	Testing process		Finances	
	Relationships		Lack of HD facilities	
	Children		Lack of understanding of	
	It's a monster		HD	
	Workplace			
	Social support			
Resources	Information	Partner and family	Cure	Adaptation over time
	Counselling		Possible facilities	
	Medication		Religion	
	Coping		Disability grant	
	Employment		Life insurance	
	Social support		Medical aid	
	Testing process			

## **4.2. Challenges**

Challenges experienced by individuals with HD were one of the focuses of the current study. It was apparent from the data analysis that the participants in this study faced several challenges. Sub-themes related to the challenges faced by these individuals were identified on four of the five system levels of Bronfenbrenner's ecological model (Bronfenbrenner, 1979). Subthemes that related to the micro-system included: triad of symptoms, testing process, relationships, children, it's a monster, workplace, and social support. There was only one challenge identified on the meso-system, which was partners and family. There were no challenges identified pertaining to the exo-system, meaning the next system was the macro-system, which included the following challenges: medical aid, insurance policies, finances, lack of HD facilities, and a lack of understanding of HD. Lastly, there were two challenges identified on the chrono-system, namely symptom watching and the progression of HD. These challenges are discussed in more detail below.

### **4.2.1. Micro-system**

The first level on which challenges was identified was the micro-system. The micro-system, according to Bronfenbrenner, includes those interactions that happen in the person's immediate environment, that which they are operating in, for example their workplace, peers and family and even their own body (Bronfenbrenner, 1979). There were several challenges, which included the triad of symptoms, the testing process, relationships, children, HD being a monster, the workplace and lastly, social support. These are discussed in the following sections.

**4.2.1.1. triad of symptoms.** The first challenge that was identified as existing in the individual with HD's micro-system was the triad of symptoms and consisted of motor disturbances, cognitive dysfunction and behavioural problems. Almost half of the participants reported motor disturbances to be a challenge to some extent. Motor disturbances that were reported included uncontrollable movements, problems with balance, slow reflex responses, difficulties with walking or problems with writing. Participants described how their bodies were constantly moving involuntarily when they were sitting down, so much so that their movements could break chairs. It seemed that the uncontrollable movements were often experienced in the participants' extremities, such as their hands and feet, as some stated:

My hands and feet constantly move. (F10) (Translated from Afrikaans)

(Points to arm)...and legs. (M11) (Translated from Afrikaans)

Neck jerks, arm ticks, constantly playing with fingers, fingers are constantly going.

My mouth is constantly like chewing something all the time. (M9)

It was evident that the participants' hands were often mentioned as an extremity that is severely affected by HD. As a result, writing can become a problem and one participant explained how illegible his writing had become:

My speaking, writing...all coming into effect. It was very poor. Could not even read my own writing. (M9)

There were also two participants who described that their walking was affected by HD. They either experienced a retardation in walking speed as one mentioned:

My walking is quite bad now...but I do walk slowly and it effects my life... (M9),

Whereas the other participant reported difficulty with his/her? co-ordination and reflexes when walking, as seen below:

I think coordination more than anything else. Well you know if you trip the sort of coordination to recover...Isn't really there, the quick reflex. *Yes* [ya] in fact that's probably what it is, it's a reflex that's missing. (F6)

Dropping objects and knocking into things were other results of uncontrollable movements and were reported by three of the participants as a challenge. One of the participants said: 'I am always moving around quite quickly and walking fast, so naturally, I drop things...' (M1), and others shared this sentiment:

It is just that I break things at home, like cups and those sorts of things. (F10)  
(Translated from Afrikaans)

I can tell you that I bump things. (F8)

Many of the motor challenges also involved the mouths and lips of the participants. Participants eight and eleven reported that they experienced speech difficulties, such as forming their words properly and they could also not manage to eat and swallow without making a mess. This was described as a challenge as these two participants explained how it was very frustrating not to be able to form their words quick enough or to even form them properly. This is illustrated by the following two excerpts:

Its more irritating because sometimes when I want to, if I want to say something in a hurry I can, it feels like I am tripping over my words. So if I want to say to Jack (pseudonym) hurry up and get done, I find it harder to find my words. I have noticed that if I try to say 's', the 's' sounds in particularly, 'silver sixth Spence', I can catch it. (F8)

My words can't come out...it's slow...and soft. Talking is very difficult for me. I can't really talk. My tongue moves. (M11) (Translated from Afrikaans)

In addition to motor disturbances, cognitive dysfunction was also identified in the participants' responses as a major challenge. It should however be noted that no formal evaluations were conducted; these challenges relate merely to the participants' perceived cognitive difficulties. The first type of cognitive problem that was noted by a quarter of the participants was their lack of concentration. This was described as a challenge that occurred at their workplace in particular. A lack of concentration lead to them making careless mistakes in their work and finding it difficult to focus on their work and in meetings. The following excerpts illustrate their concerns:

The company didn't continue my contract because of my lack of concentration. They basically said that I was making silly mistakes, so, with this psychiatrist there was a little test that I did and I was showing some signs of ADHD. (M1)

At work I can focus on things only for a little while...But normally what happens is if I'm I've got a shorter attention span I mean, I sort of find myself like drifting and not focusing. (F2)

So sometimes like I'll be in a meeting and somebody would be like what do you think. I'm like oh God, what was it? You know I just space out....so but yes [ya] like definitely lack of concentration. (F3)

Concentration problems generally seem to get worse as the day progressed. The participants reported that towards the end of the day, they felt that the lack of concentration was more intense. This problem is illustrated below:

It wasn't as much the type of activity, but usually the time of day. From about 2pm I really struggled to focus on the work I was doing. (M1)

It was more towards the end of the day...as I got tired. (M9)

Concentration problems were however not only experienced at work, but also while driving. Two of the participants stated how their concentration problems affected their driving ability, especially when they have to travel long distances, as one of them stated:

What I do think I will battle with is a long drive. (F2)

Another participant explained she has been reminded by her daughter to be attentive while driving as her mind tends to wonder off:

Be more alert...I know my daughter has said to me on a couple of occasions mom you must keep your eyes more wide open. (F6)

One participant explained that if she had to stop driving because of HD so as not to put her family in danger, then alternative arrangements for transport would have to be made, such as hiring an au pair. However, this is a very expensive alternative, as she stated:

I don't want to be dogmatic about driving and about putting myself and the kids in danger...I would have to have an au pair to drive around, but now we are looking at R5000 a month and we can't afford that. (F8)

Another problem that was reported by the participants, as a cognitive problem, was that their memory seemed to be affected by HD. In some cases, memory problems made them forget their personal belongings, such as their wallet at home when they went to the shops, or they would forget which word they wanted to use. One participant also reported how his

memory problems negatively affected his work. These issues are conveyed in the extracts below:

I cannot remember things well anymore. And I have put things away like my wallet and when I got to the mall, I realised I forgot my wallet en then I had to turn around...the drawback from it, the forgetting...I mostly don't remember anything.

(F10) (Translated from Afrikaans)

*Yes [Ya]*, my memory started being affected and a lot of stress with a management job and so I would very slow the fact that I'm getting slower and slower. (M9)

Uhm, I always forget I am trying to say at the end of a sentence. So I'll try to say, James (pseudonym) go get the...whatever...in the car, I can't remember where it is....the jumper in the car... I can't find a word sometimes. (F8)

One participant also reported that the cognitive symptoms of HD were the most frightening of the condition. This was because the individual felt that it would be impossible to acquire new information or to assess situations, or even to help family members while severe cognitive deficits were experienced. This sentiment is expressed in the following extract:

For me, the worse side of Huntington's is the mind... so I am worried about not being able to take in information, or to be able to analyse things, not being able to cope, or help my daughter in a situation. I think it is memory loss. I can handle all the other HD related symptoms, but for me, the loss of cognitive function, that is a big worrying thing. (F5)

The last of the triad of symptoms that emerged from the data analysis are behavioural or psychological problems. It was evident in the responses of half of the participants that

depression or feelings of sadness were experienced. This was often related to their own positive test result of HD or when other family members were diagnosed with HD. Some responses included:

*Yes [Ya] it was a bad time for me but yes [ya]... what I can say is I still haven't recovered from my depression and anxiety from my results. (F2)*

*Yes I did. I picked up a lot of weight. I started comfort eating. What I tend to do is I work on auto pilot during a crises, so during the time I was ill I did what needed to be done, and I think when she passed away, and it was actually quiet, that's when it hit me. ...I did go through a low dark time. (F5)*

*I have been (depressed) previously yes, especially at the stage when my father was diagnosed, especially as a family we buckled... (M12) (Translated from Afrikaans)*

Other psychological problems that were noted in the participants' responses were self-esteem issues. These related for example to feelings of embarrassment when the uncontrollable movements were getting worse. Not only was a problem with self-esteem raised, but another participant also described how she experienced a lot of anger. These are evident in the following excerpts:

*...self-esteem issue because I am more conscious about my body and I can't do what I want to do and my body going all over the place. (M1)*

*My friends say I am more relaxed. I have stopped taking them (medications). But then they said I was like getting angry....So apparently that is also part of the mood swings. (F8)*



**4.2.1.2. *sleep problems.*** Another challenge that was reported by several participants was regarding sleep. Almost all of the participants reported sleep disturbances, which included falling asleep very quickly, being unable to fall asleep, taking long to wake up, as well as experiencing increased movement either just before falling asleep or during sleep. The first challenge that was reported as a sleep problem was that participants fell asleep very quickly. These sentiments are described below:

...generally I can't even read a book....cause I just climb into bed and pass out...Five minutes...Head hits the pillow and I'm gone *yes* [ya]. (F3)

Oh probably five minutes...I fall asleep very quickly. (F6)

Another problem, on the other spectrum of falling asleep quickly, was that a third of the participants struggled to fall asleep. This was partly attributed to stress after receiving their HD results and having to think about the future, as one participant explained how he would lie awake and stress about how he was going to provide financially for his family when his symptoms progressed. Another participant stated:

I have sleeping problems in the beginning when I had the diagnosis...I would just not be able to go to sleep. It's one of those when you are awake till 2 in the morning and then you have to get up at 5 and you are still grumpy because you are tired but you actually cannot switch off to sleep. (F5)

Two more participants reported that they could not sleep for very long and would wake up after a short period of sleeping or would wake up frequently during the night. This is reported below:

I can't sleep. I take very long to fall asleep. (F10) (Translated from Afrikaans)

I don't sleep. I only sleep two hours a day...I don't sleep...I'm awake the whole night.

(M11) (Translated from Afrikaans)

One participant also reported that she recently felt that she took a longer time to wake up and that this often made her late for work.

Lastly, it was also reported by two participants that they experienced increased involuntary movement, as one of them reported that he felt restless and experienced sudden jerks while sleeping. Another participant experienced movement just before she fell asleep and responded:

Just reasonable amount of movement and stuff but once fast asleep it's fine...Just when falling asleep there's movement. (F6)

**4.2.1.3. testing process.** The next challenge that was reported by the participants was the process of predictive testing. Predictive testing is when blood is drawn from the individual who is at risk of HD and analysed to establish their CAG counts in order to determine whether they would develop HD or not in the future (Gudesblatt & Tarsy, 2011; Huniche, 2011; Roos; 2010) [see detailed discussion in literature review in the diagnosis and treatment section]. For the participants, the testing process involved the reasons behind having the predictive test done, medical staff who were involved, long periods of waiting, travel distances, and lastly, the participants' reactions towards the test results. All 12 of the participants in this study went for predictive testing, as a confirmed diagnosis of HD was one of the criteria to take part in the study. The first part of the testing process involved the decision whether to go for the predictive testing or not, because many major life choices seemed to be dependent on the results. One of the reasons why two of the participants wanted

to have to go for the predictive testing was to decide whether they could have children through normal conception or whether they would need to seek medical help, such as in vitro.

I got married about 3 years ago, so we decided to rather get tested, because obviously we wanted to know regarding getting kids, you know the various options we could take after that. (M1)

I had the onset of marriage and that I wanted to have babies...after the predictive testing...I wanted to know because my next would have been to uhm, there is a certain IVF that... so they would have fertilised the right one. (F4)

Whereas some participants wanted the test to help them to make a decision regarding which method of conception to use, others had the predictive testing done in order to know if they wanted to have children at all, and this decision seemed to depend on the predictive test results. This is described in the following excerpt:

Well basically, I was in a relationship with someone and was at the stage where it was like do I have to get married to this person; whether or not I can have children. (F2)

Another participant reported that she wanted to go through the predictive testing because she wanted to know whether her children were at risk of having HD, as she could have possibly passed the HD gene onto her children. She said the following:

...so that was the main purpose... I was hoping that you know, that I wouldn't be positive and then we didn't have to worry beyond that. (F7)

There were also some challenges described that related to the medical staff who were involved during the predictive testing. Many of the participants felt that there were too many specialists involved because they had to see a psychologist, psychiatrist and a neurologist.

Several of the participants also felt that some of the appointments with the specialists were either unnecessary or not thorough enough. Some of these challenges are demonstrated in the following excerpt:

...they make you see a psychiatrist, a psychologist and a neurologist. So you have to go through all that testing before they will do the blood tests. To me it was almost as if people have a set idea of how HD should be and how HD people should act. So, like to me, the psychiatrist and psychologist was a bit unnecessary. My experience of the psychologist was not good. We had an hour session with her where she literally just toppled the basket and sent us home so you know, so once she unveils all the ugly and then sends you home because one session is all that is required for the testing.

(F5)

Some of the tests and procedures were also not explained to the participants by the medical specialists. Participants felt that they did not know why certain tests were being performed on them and reported that the medical staff were unwilling to inform them when they enquired. This is evident in the next quotation:

I saw a lot of people. And they couldn't tell me why the stuff was being done. We just felt, if you go for a test like for cancer or that, you know, they do the blood tests and say sorry, you have it, but with us, we had to see neurologists and specialists just to get the same result to say what exactly is wrong with you...and what also caught us off guard, are the neuropsychological tests that they did on us, and I don't really understand why it was done and they also couldn't tell us why it was being done.

(M12) (Translated from Afrikaans)

Another challenge regarding the testing process was the time it took from when the participants started the process until they eventually received their results. This was described as a lengthy process by a third of the participants, as there were many steps involved and long periods of waiting. Some of the participants wanted to receive their results sooner as they felt they wanted to deal with the outcome. These challenges are expressed in some of the participants' responses:

They did, it was, if I can remember, it took like 2 years. It took very long, backwards and forwards, backwards and forward, from the one month to the next to the next.

(F4)

So I found it very very long and some of it very unnecessary... And the 6 week waiting period to get your results were excruciating. So in between all of that, I think it was a horrible, I think everybody just wants to know. You want to get to a point where you can say either yes I do have it or no I don't. But for me personally, the waiting was excruciating. It's that what if thoughts that you play through in your mind all the time. (F5)

Very stretched out...it was an incredibly stretched out process. (M12) (Translated from Afrikaans)

In addition to problems with medical staff and long periods of waiting, some of the participants also had to travel quite a distance for all the appointments that were necessary for the predictive testing. The following sentiments convey this challenge:

I lived in Pretoria and I had to trek all the way to Johannesburg to have all those testing and stuff. To go to Sterkfontein mental hospital and these sort of things and then I had to go to Wits for other stuff so it was a lot of travelling. (F7)

It was really difficult, we had to drive far...(M12) (Translated from Afrikaans)

Lastly, the participants' reaction towards their positive HD result was also considered to be a challenge for many, as many of them were shocked when they received the news. According to the participants they were shocked because they never considered the idea that they would in fact test positive for HD; in their minds they were always negative for the HD gene. This is described below by the participants:

By the time we got the test then, I mean, I'm such a positive person, so in my mind I was convinced I'm negative and everything else, but as soon as she said I'm sorry, but it's positive so it's completely heart breaking. (M1)

Was pretty bad...it was terrible because my brother had tested positive. My father tested positive. My father's got three sisters, so my three aunts all went to get tested for predictive testing and they all tested positive. So there's like sixteen of us cousins at risk and essentially every single person that got tested has got a negative, has got a not a good result...I think we both really just wanted to be able to change it because...everybody gets tested it's negative, so they also maybe don't wanna get tested, so I think it was important to have someone who wasn't (positive)...to kind of break that cycle. So I think it was a lot depending on it and it didn't work out the way I hoped. (F3)

So both my sisters and I decided that we'd go off and be tested, not thinking for a minute that we would actually be tested positive. We thought well Patrick (pseudonym) and dad had been male...so we're definitely going to be negative...yes [ya] it was just a terrible shock to everyone I think...I was in shock. I was so positive I was going to be negative. (F6)

The test results left some of the participants feeling devastated, as they were not prepared for the results, as one of them said:

Shocked. Disappointed. You don't really expect that sort of a result. (M12)

(Translated from Afrikaans)

Other participants felt that they let their families down by receiving a positive result for their predictive test because they did not want their children and spouses to experience the burden of HD. These sentiments are conveyed in the following extracts:

I wanted to know, right, that is the end of that because of the children...we just wanted it to stop there...disappointed for the kids more than for myself...I think disappointed because I had hoped it would end there. (F7)

The first thing I said was I said sorry to my husband because I don't think that is what he had in his plans for his future and I started crying. To me it just felt like I had disappointed him by getting a positive result. (F5)

Other reactions from participants included that the outcome of the results felt unreal. Some described it as a dream-like experience and a few days were needed to completely process the results, as one participant said:

I was really shattered...a good three to four weeks I couldn't believe it. I kept thinking that it's a dream. I'm going to wake up and it's all going to go away and it's not going to happen. (F6)

Others shared this sentiment and reported:

...but after a day or two it really sinks in, the reality of it of what is going on and then it hits you what is waiting for you, especially now that I see my dad, it's pretty scary.

(M12) (Translated from Afrikaans)

I must say it really did affect me. When I was told, I think I went numb and I did not break down and cry because I don't think I actually like took it in. (F2)

Others felt frustration as they pondered why it had to happen to them and they felt that the positive outcome of the results was undeserved. These sentiments are expressed in the following extracts:

It's unfair...Its *yes* [ya] kind of blows your mind a bit. (F2)

I just felt *yes* [ya] frustrated dammit. Why has it happened to me? (F6)

Lastly, the emotional distress and devastation that resulted from the positive outcome of the test results are evident in the following quotations:

Slightly more fragile....Not a whole human being; all of a sudden half of you doesn't work. (F5)

It was hectic...I spent a couple of days crying in the cupboard...It's very overwhelming.

It is a very scary thing to face. (F8)

**4.2.1.4. relationships.** Another challenge that was expressed by the participants was the effects that their test results had on their existing or future relationships with their partner. The first challenge that was reported was that some relationships unfortunately ended shortly after some participants received their results, as shown in the following excerpts:



So you can decide if you wanna do this or not...he was like okay fine and then we started the testing process and then he actually just, he broke up with me but I don't know...he sort of said it didn't really have anything to do with that but I have my reservations.(F3)

The result was that the relationship deteriorated even more. So I'm no longer in contact with that particular person. (F2)

Another challenge related to being in a relationship and knowing you have HD was that participants were hesitant to invest all of their energy in the relationship, because they were scared that their partner might not want to get married because of their HD diagnosis. This sentiment is expressed below:

...it's a bit difficult because I kind of feel... I haven't got my heart like a 100% set on the fact that we like, like if I think about marrying. I will, obviously I wanna marry him but I don't, like I've never said it to him like that because I don't wanna put pressure on and I think also maybe just I've tried to keep a bit of a...so if it doesn't work then or if he says look this isn't my life journey then I haven't sort of you know given it my all... (F3)

Entering a new relationship was also reported as a challenge by some, as some participants said that they felt that they would be self-centred to enter a relationship, knowing they might have or have HD. Others felt that they would put extra pressure on the relationship when they shared that they have HD. These concerns are stated below:

...well the irony was, we actually started dating the day after my father's funeral. No, I mean it was...it was tricky. (M1)

Whether or not I'm going to actually be involved with someone, it's a very difficult decision because once again I do not want to be selfish to that person....but if for whatever reason it does get serious I would have to disclose to them and then it's for

them to make that decision but I think on the outset of relationships I also say listen you know what, I'm not interested in getting married and definitely no children. I don't want to put that pressure on the person. (F2)

Entering a new relationship was also reported as a challenge as the person with HD would have to tell their future spouse that because of having HD, normal conception would not be possible and medical procedures would be needed. Other methods of conception included in vitro fertilisation, to ensure that the fertilised egg will not have the HD gene, meaning the child would not be at risk for HD one day. This challenge is stated in the quotation below:

I am careful of that one (entering a relationship). For me it is actually a difficult one because I have to tell my wife one day, listen here, I am sorry but I have this disease, I can't have children normally, we will have to get medical help to have children one day. So there are things that will bother the person... (M12) (Translated from Afrikaans)

**4.2.1.5. children.** In some relationships, the decision of whether to have children or whether to adopt a child was also considered a challenge for the participants. The first challenge that related to this theme was the consideration of alternative methods of conception, as to not pass on the mutant gene. Methods such as in vitro fertilisation are stressful and very expensive. Adoption was also often under consideration. These struggles are conveyed in the responses below:

I think, originally, obviously the cost is a big factor....but I just think from our point of view, we were so scared of what to do, what to do, and one day we just decided to have a baby the normal way. (M1)

...I always thought that you know now if I wanna have babies am I going maybe adopt because I don't wanna pass this on... yes [ya] I mean in vitro is also quite...quite stressful and if it doesn't work and it costs a lot of money... (F3)

Ok that was a big thing. I wasn't going to have a child. I wanted one, I would have had 10 if I tested negative, but it was positive so then I, I was going to do the cell in vitro fertilisation, but where they do this thing for Huntington's...but it wasn't ready, it went on like for two, three years...so my husband said, you know what, let's put our faith in God and then we moved on. We had our son. (F4)

Other participants even went so far as to say that they did not want children anymore after they learnt that they tested positive for HD, as one participant stated:

I'm not interested in getting married and definitely no children. (F2)

Some of participants that already had children felt guilty about the possibility of passing the HD gene on, as some of the children had their own children already or still needed to find a partner that would be accepting of HD. The excerpts below convey these messages:

So, it was, yes [ya] it was an uncomfortable feeling that we couldn't say right, that is the end of that because of the children...but I am more concerned about my kids because it's a big burden for them. They already have these 2 children but obviously he just worries about them because if he is a carrier then the chances of them having it also...Then our daughter, because she hasn't found a partner whom she knows is accepting or whatever, she still I think it just plays on her mind a lot. (F7)

The last challenge that was mentioned about children was the difficult decision to terminate a pregnancy, as the couple did not want their child to have HD. This decision was

further fuelled by the slim chance of a possible cure in the near future. This challenge is depicted below:

That was the worst thing ever *yes* [ya] like it was awful...There's no words you can't describe it that is the worst point....because there was no cure and nothing on the horizon looking good and for a cure. They recommended it you know very strongly that you terminate. That was one of the darkest days of my life and I know that because it wasn't a planned pregnancy it was an accident. (M9)

**4.2.1.6. *it is a monster.*** Half of the participants expressed intense and strong emotions around HD. The disease was equated to a 'monster' (M1) and a 'curse' (F4). These sentiments convey the immense fear and intense loathing that the participants felt towards HD, as others also described it as:

It's there; it's ugly it stares you in the face, it's there. (F4)

At times you kind of, it can, if you let it, overwhelm you, but other times you don't. (F8)

I don't want it. (M11) (Translated from Afrikaans)

All of these sentiments show that HD can be a very frightening condition to be diagnosed with, and evoke intense feelings of anxiety in some participants.

**4.2.1.7. *employment.*** HD also affected the participants' work performance. This was related to the variety of consequences related to HD, such as the memory and concentration problems that were discussed earlier. Some of the participants mentioned that their work contracts were not renewed, due to HD related problems, as one said:

And then my next job after that they didn't extend my contract too, just purely because of lack of performance. So obviously, from my point of view, it was hugely, hugely hurtful. (M1)

Others had to stop work as a result of HD as mentioned below:

I had to stop working because of the HD last year now. *Yes* [Ya], my memory started being affected and a lot of stress with a management job and the fact that I'm getting slower and slower. (M9)

Then I couldn't pick up things anymore, and then I couldn't work... (M11)

(Translated from Afrikaans)

Whether the challenges related to a contract that has not being extended or having to look for other work or simply not being able to work at all, it was evident that most of the participants experienced work-related challenges. This lead to stress and anxiety, while others had to face a financial loss. These challenges can be seen in the following excerpts:

...my sleep problems were work induced, I wasn't actually working. I was actually at the point where I was unemployed and was job hunting, and the added stress and anxiety...so stress and anxiety, I think, made my sleeping pattern a bit crazy. (M1)

Overall, I've missed out on you know quite a lot of money...Bonuses, I don't get access to profit shares, nothing like that anymore. Profit share. I do get my basic salary as a pension you know that type of thing, but it does not cover everything. (M9)

A last challenge that was mentioned as a workplace-related difficulty was the fact that while they were working full-time, some individuals had to deal with the positive outcome of their test results as well. This was emotionally taxing for some and gave them little time to

process their results. This was particularly challenging when there was no support from the employer, and no willingness to make special exceptions. This is conveyed below:

But the emotion was basically it's difficult especially working. I work five days a week and basically still trying to cope with life and to be honest, I was not coping...And especially with the large workload and then I get this test result. And it's kind of hectic...At the moment, there's really no support for this type of disease at work and it's like, for example, you can go to the employer and there's a program that will assist you, like for example they've got HIV Aids they'll be able to assist...They can place you for example like a half-day job, or in a place where you'd be doing less work. But at the moment, there's no support from the employer. (F2)

It is evident that the participants faced several work-related challenges because of HD and sometimes the disease led to losing their jobs. Three participants had to stop working because their HD had progressed too far. HD therefore has the potential to interfere significantly with an individual's work circumstances.

**4.2.1.8. social support.** Social support, in terms of the participants' partners or spouses, extended family and support group, was also sometimes referred to as a challenge. One participant reported that her spouse could be unsympathetic and intolerant at times. Instead of her husband being impatient and dismissive towards her, she would prefer to feel that he still sees her as a valuable asset to the family. This challenge is described below:

Ryan (pseudonym) can dismiss me fast. He can say at the moment, I am very hard to deal with or I am very hard to understand...He will cut me off and say, I can't understand you or you know, you are so hard to understand, or so difficult to understand, so he cuts me off without giving me the space to say something...I am still

understanding, I am still like part of the plot and that is hard for him to sometimes realise. (F8)

Another participant mentioned how his wife got irritated when they went for walks and he could not walk as fast, as he stated:

I do walk slowly and...it bugs my wife. (M9)

Not only were spouses mentioned as a social support challenge to the participants of this study, but their families were as well. Challenges that related to the participants' families included: having false perceptions about family members with HD, disclosing their positive test results to the family, difficulties and concerns about family members with HD, denial of HD and participants viewing themselves as a burden to their families.

The first challenge regarding the participants' family was that some of them had false perceptions about, or had misjudged, family members' behaviour or movements because no one knew about HD. One of the participants even mentioned how his family thought a family member was an alcoholic because of her strange movements and that they did not realise that she had in fact been showing signs of HD. Another participant explained how she used to criticise her own mother because the participant also did not know her mom had HD, as she said:

I think growing up it was very difficult to understand why she did the things she did and we only realised once we got the diagnosis and we understood what the disease was about; why she did the things she did. So in hind sight I think knowing would have actually helped us better understand where she was coming from...I just feel that I might not have judged as harshly. (F5)

Another challenge that surfaced regarding the participants and their families was about disclosing to the family that they had tested positive for the HD mutant gene. This was a difficult task to do as one said: 'Oh, it was terrible *yes* [ya]...', (F6), as sometimes they did not expect to get the results that they did or some had to hide their results from their family members, such as their children, as a way to protect them .. The following excerpts show these difficulties:

I haven't told my daughter yet. She is 14 at this stage and she can't go for testing...she can't go for counselling, and she can't do anything with it. So we have made a decision where we actually got to a point after the testing that we won't tell her (about mother's results) in time. So it is not that we want to hide it, it is just that she can't do anything with the information so when she is 18 she can go out and get tested herself. (F5)

Oh, it was terrible *yes* [ya]. No it was hard telling the family. And I think because I sort of had built up in my own mind that it was going to be negative so you know. (F6)

HD is a hereditary condition and is often present in more than one member of a family. Often children with HD end up looking after a parent with HD. So it was not surprising that a number of the participants found dealing with family members with HD challenging. Family members were viewed as social support in this study, and as this challenge also pertains to family members directly, it was considered also to be part of social support. There were four main reasons, which were: having to do a lot for the family members with HD, seeing them suffer, HD being kept as a family secret, and being scared by seeing family members with HD. The first reason why other family members with HD were considered to be a challenge was because the participants had to do a lot for them. Tasks that



participants had to do for their family members included grocery shopping, driving around, organising nursing care as well as getting their medication for them. Some of these challenges are expressed below:

I was always the one sorting things out, and checking pills, sorting nurses, getting her to the hospital. (F5)

And then my mom spent 4 years here in the house and then Groote Schuur came to me and said you can't cope any further... I can't look after my mother by myself, it's very nice, it's very noble, I cannot do it...I could never take her to Pick 'n Pay; I could never take her to the bank. I had to do everything for her. (F8)

The second challenge with having to deal with family members with HD was that many families tried to hide the disease from others, almost like a family secret. My impression was that the families were embarrassed of these individuals with HD and did not want others to know about them. These concerns are expressed below:

...it was swept under the carpet, you were hidden away in the back room somewhere, it was never spoken about, it was hidden away...So it was hidden away and I didn't want that. (F4)

Well basically if we speak about the Huntington's, we never really knew what my mother had, growing up. And I think back in the day they used to hide everything under the carpet; it was a big hush-hush thing for anyone who had an illness. (F5)

Some sort of murmurs, someone had something strange somewhere, but something strange is neither here nor there. (F8)

Another concern of the participants about their family members with HD was that they had to watch them suffer as the disease progressed. This was mentioned by most of the participants as all of them had other family members with HD. One of the participants explained how it was difficult to see her brother struggle financially, as he was battling to find a job and there was nothing that she could do for him. Another participant experienced a similar situation and explained how some of her family members who had HD ended up living on the street because there was no financial planning done within the family and it was heart-breaking for her to see them struggle. The same participant also said that her mother had become a 'burden' for their family as she could not eat or swallow properly and she found it very difficult to see her mother regress to that stage.

The suffering of the disease was emphasised as HD can progress for up to more than a decade, with slow deterioration. One of the participants explained: 'it's not two weeks or 2 months and then you are dead...it could be 15-20 years' (F4). This slow deterioration was difficult for some as they had to watch loved ones slowly regress more and more, as this participant described it:

Now my mom lingered, so my mother did the long years of lingering, you know, which is different. You know what you can watch, the going downhill, or the drama, you can watch. It's different. It's harder; it's harder on you, than someone that dies suddenly. (F8)

One of the participants reported that she felt terrified to see her brother struggle with the disease because he got worse as time went by: '...to see how he deteriorated was very frightening...' (F7).

Another challenge that emerged with regard to family members with HD, was that the participants got a glimpse of what their future might be like. This was described as a challenge as many of the participants were frightened about their future. One of the participants reported:

...but after a day or two it really sinks in, the reality of it of what is going on and then it hits you what is waiting for you, especially now that I see my dad, it's pretty scary.  
(M12) (Translated from Afrikaans)

This sentiment was shared amongst the participants and is evident in the following quotations:

And my brother has also made it very real to me how soon it actually can kick in. (F3)

To go and visit everyone so it's very, very difficult. They are far worse than me. My one aunty she's already in hospice care and that is very difficult. That is our future that we looking at. (M9)

Some participants also reported how their family was in denial about HD. This refusal to accept HD as part of the family made it difficult for others in the family to come to terms with HD whereas in other families it resulted in family disputes. The following excerpts display these difficulties:

...well she (mother) didn't want to accept to start off. It took a long time to realise you know if people bumped into her and saw Patrick (pseudonym) walking down the road, was all they would say to her, you know, 'what's the matter with Patrick?' Oh no you know he's just having a couple of bad days. She would actually never admit to the fact until one day someone said to her Joan (pseudonym) you know you actually have to admit what's happening. You can't hide it any longer. Patrick's just going to get

more and more you know ill and people are going to realise that and I think once my mom came to terms with it all, not came to terms with it but I think once she accepted it then I think it was easier for everyone because at least then we could actually talk more openly about it but it took her a long time to accept it *yes [ya]*. (F6)

Family wise, people don't really like to talk about it. I think they are still in the denial phase. That would be my side; my dad, my brother and my sister. There is a lot of family bantering that goes around with regards to the Huntington's. (F5)

The last challenge regarding the participants' families is that a third of the participants viewed themselves as being a burden to the family because they have HD. One of these participants reported how he had to rely on his family because he was not working anymore and thus spending more time at home. Another participant explained how her mother looked after her and assisted her with getting on the taxi for example, but then stressed the whole day about her. In the words of two other participants:

I said to my doctor you just inject me with some kind of thing and leave me because I don't want to become a burden...that's a big thing. Because you become a burden, you can't eat and drink and you can't swallow and everything is messy and... (F4)

Ya, and also on top of that, it is a double edge sword. You don't want to be a burden to those you love, but I also don't want to be in an old age home. (F5)

The last form of social support that was mentioned as a challenge related to the support group for individuals with HD. These support groups are held once a month and run for about 90 minutes, with the occasional professional speaker, such as a genetic counsellor, that addresses the group. Although the participants in general found the support group a great source of support, some of the participants reported that the emotional tone and content of the

support group could be tiring and upsetting. One participant reported that they have to deal with the group's emotional turmoil in addition to their own. This dilemma is described below:

I don't want to know about how you (group members) are feeling, because that's different...It's just me personally. I think I am trying to deal with my own bloody crap here and I don't need to hear theirs. (F8)

#### **4.2.2. Meso-system**

The second system of Bronfenbrenner's ecological model is termed the meso-system. This system comprises of the interactions between the micro-systems, for example the interactions between the participants' families and their friends (Bronfenbrenner, 1979). Only one challenge emerged on this level and related to the interaction between the participants' partners and their family.

**4.2.2.1. *partners and family members with HD.*** Only one participant mentioned that it was difficult that her partner observed and met with some of her family members with HD. According to her, this was potentially challenging for him because it showed her partner what could possibly happen to her in the future if her HD progressed to that stage. This challenge is expressed below:

When he (brother) was down here, it makes it real to Hayden (pseudonym). So I think to him it makes it real you know, that's how I could be and that, yes [ya], so that's a challenge. (F3)

#### 4.2.3. Macro-system

No challenges emerged on the exo-system. This is because the exo-system consists of links between the micro-system but where the individual is not actively involved, for example the participant's partner and their partner's workplace (Bronfenbrenner, 1979). As the study focussed on the unique individual experiences of the participants, there were no challenges identified on this level. Therefore the next level where challenges were identified was on the macro-system. The macro-system consists of the individual's culture and values, as well as encompassing the laws and policy making which affects the individual (Bronfenbrenner, 1979). Several challenges were identified on this level and included: medical aid, life insurance policies, financial problems, a lack of HD facilities and a lack of understanding of HD.

**4.2.3.1. medical aid.** The first challenge that emerged on this level was medical aid schemes and the problems they posed. Almost half of the participants stated during the interviews that they had a private medical aid scheme. However, four of the participants reported that they felt frustrated with their medical aids. According to the participants, they have to be on the most expensive and comprehensive plan because they are diagnosed with HD, however, there are not a lot of medications available to treat HD. Some of their frustrations are illustrated in the responses that follow:

Discovery is almost R3500 for a main member if you want to go on the top plan that covers Huntington's. The problem with Huntington's is that it is not on the prescribed minimum benefits list, so Huntington's is one of those that is excluded, with regards to medical aids. There are 3 plans that will pay for Huntington's but ...you also have to go to the top plan of that specific scheme to be covered so expense wise, it is a lot more expensive to put these things in place if you do disclose your status. (F5)

Discovery Health, the medical aid, If I had to tell them I had Huntington's, you can only sit on the top bracket. They don't let you sit on anything else, you know what I mean, to cover me. (F8)

**4.2.3.2. life insurance.** The second challenge that emerged on this level related to life insurance. This was described as a challenge because it was difficult to acquire life insurance as some were rejected a few times by insurance companies once the outcome of their predictive test results were positive for HD. One participant said:

Obviously stuff like...life insurance, because it's in the family, we have got ourselves organised years ago...we tried so many different companies...and long time ago when we were starting to apply, a lot of them said no...(M1)

Two of the participants described insurance policies as complicated. According to these participants it is best to have a policy in place before you decide to go for the predictive testing. These sentiments are described below:

He said to me if you decide... to get tested just know that you need to have all your cover in place first... (F3)

...there are a lot of loop holes and you can get yourself into big trouble if you don't know, you know, you need to know what you are on about. (F8)

Two of the participants also reported that because the outcome of their predictive test results were positive for HD and they disclosed their results to the life insurance companies, life insurance was very expensive. This is described in the following quotations:

Like I'm poor in the month for the amount of money I put in this policies I don't know if it's going, I don't wanna have to and also *yes* [ya] I don't wanna have to worry about money. (F3)

Life insurance becomes more expensive. Anything is double the price if you have had predictive testing because if you don't disclose that and you end up with a degenerative disease, you don't get paid out so what is the point of paying insurance all your life because you are not going to get anything out unless you have disclosed and filled out forms saying I have had predictive testing. (F4)

**4.2.3.3. *financial problems.*** Financial difficulties due to HD also emerged as a major challenge. From the 12 participants who were interviewed in this study, 3 had to stop working because of HD, and as a result experienced financial difficulties. This was evident when one participant explained that her only income was the grant that she received: 'Only from Groote Schuur, they gave me the grant.' (F10) Another participant explained how he did not receive the financial benefits from his company anymore and that his monthly income had decreased:

Overall, I've missed out on you know quite a lot of money...Bonuses, I don't get access to profit shares, nothing like that anymore. Profit share. I do get my basic salary as a pension you know that type of thing, but it does not cover everything. (M9)

Another participant mentioned how that as a child, financial difficulties were imminent as her mother had to work to pay for nursing care in order to take care of the participant's father who had HD, while others reported that they could not spend their money frivolously but had to save as much as possible to provide for their family's future. The following excerpts describe these sentiments:



So yes [ya] like put a decent amount of money away every month and you know my mom was a teacher and she had to work, so many extra lessons just to pay for the nursing care and you know...it was always a struggle financially and was always tight...if I'm sick the last thing I want my husband to worry about are finances...(F3)

I think a lot care-free. I didn't have to think 3 times about everything before I did it...I would not buy a car now, I would much rather put my money in the bank to look after the family when I am no longer part of the unit. So you turn around every cent 6 times before you spend it. (F5)

**4.2.3.4. lack of HD facilities.** In addition to medical aid, life insurance and financial problems, participants also had to deal with a lack of care facilities, whether for themselves or family members with HD. The issue of not having a proper care facility, which has professional staff who are trained to care for individuals with HD, was raised by a few participants and was considered to be a serious issue. One of the participants (M9) explained how it was appalling to see the amount of care and care facilities that other countries, such as England, offer for individuals with HD compared to the lack of facilities and care available in South Africa and how he is left to provide for his own care. Some of the other participants' responses are shown below:

...yes [ya] it's terrible. Ag it's just very, it's just sad you know and if they do go somewhere then they might as well not go because they do not receive proper care. The staff is usually not trained for people with HD. (F3)

In South Africa we do not have caring facilities for Huntington's and that is a big problem especially in the previous disadvantaged...My mom and her one sibling ended up in a half decent nice place in Stellenbosch... an old age home... staff are not

trained and geared for Huntington's sufferers...they are an old age home. So anything other than that they are not equipped for. Then one aunt ended up in Valkenberg and that was terrible because they were also not geared for Huntington's because she is not mad. Two of them ended up in a place in Mitchells Plain in Beacon Valley some like rehabilitation centre...also not geared. (F4)

This lack of care facilities that are equipped for individuals for HD, in terms of staff and equipment, left individuals feeling worried and anxious about their future, as expressed below:

Nervous, I think well you know. (F7)

...makes it very, very a scary situation for the future. (M9)

**4.2.3.5. lack of understanding of HD.** Participants also reported that they find it challenging that very few people know what HD is. This lack of knowledge may hinder the public's understanding of the symptoms that individuals with HD present with as the disease progresses and leaves individuals with HD feeling isolated and lonely. This is evident in the quotations that follow:

...I know that people shouldn't discriminate but because I think we're human in nature. People would judge you for whatever they're going to say and the thing is, especially if I have to start showing symptoms of this and there could be a stage where I might have full blown symptoms and then they won't know what it is...I don't think they understand. (F2)

...like in the beginning when nobody kind of knew about it, it's very scary and it's quite difficult to explain what it, what it was and you kind of feel quite alone. (F3)

Before I knew it was Huntington's disease, even when I did know its Huntington's disease, it didn't make a difference 'cause nobody else knew. (F4)

I just wish I could get it out to more of my friends and things. Just because I think it is good they try informing people better. I think a lot of people have absolutely no idea what it is. (F7)

Not only was the general public not aware of HD, it was even said that doctors and nurses were not familiar with the disease when they had to provide care and services for individuals with HD. As a consequence of their lack of knowledge regarding HD, proper care was not always given to these individuals. This is described below:

I think the biggest challenge that I found was that people are very ignorant about this huge disease, and that is from doctors to nurses to nursing homes...all of that and I think not everyone knows about Huntington's...I found myself time and time again explaining to a doctor why she was doing certain things...And then also towards the end, she wasn't really eating a lot and so what they did the one day they gave her soup and they fed her and one of the people who was sitting across the table told me the next day when I got in there that she actually had soup coming out her nose because it couldn't go down her throat anymore and they just kept forcing her. (F5)

A lack of knowledge and understanding about HD even extended to participants' family members (about other family members with HD) who did not recognise the signs of HD or who had ever seen someone with HD, and as a result, left them baffled and ill-prepared. These sentiments are described in the following extracts:

Whereas with Joan and my brother Patrick, poor Joan and none of us knew what was going on. He was just behaving with all these jerky movement and things and Joan had nobody to turn to and we didn't know and all of us were completely at sea. (F7)

So we sat that out and waited and then when we got the result, which was Huntington's, now my mom at that point was the only one who showed it. I had never seen it; I had no clue with what we were dealing with. (F8)

#### **4.2.4. Chrono-system.**

The last level in Bronfenbrenner's ecological theory is the chrono-system, which encompasses the changes or consistency that happen over time in the person's characteristics or his/her environment, e.g. moving houses and changes in health (Bronfenbrenner, 1979). Two challenges, namely symptom watching and the progression of HD emerged on this level.

**4.2.4.1. *symptom watching*.** The participants in this study reported that because HD is a hereditary condition and often present in their families, they would constantly analyse every movement and often mistaken normal clumsiness as a sign that HD has become manifest. One of the participants explained how she was constantly worried that when she tripped or dropped something that this indicated the onset of HD and it seemed that she was constantly monitoring her movements. Hereafter are several quotations from the other participants illustrating this predicament:

...obviously growing up, the hardest thing was, because I was my father's son... I was naturally a very clumsy person. Even if you knock something over, you automatically think you have got Huntington's...genetically, I'm my father's son, I would move exactly like him...it took me a long time to differentiate what is due to genes and what is due to Huntington's. (M1)

I think it's because it becomes a psychological thing you know when I feel my feet moving or I feel my toes or I drop something and then up there we go...but I think you know it's in my own mind that I've got to deal with... (F6)

Obviously when I trip or do something stupid, here we go, little bit of Huntington's bogging its nose in. (F7)

**4.2.4.2. progression of HD.** The progression of HD was reported as a challenge for several reasons. Firstly, the participants reported that the symptoms would become worse as time passes and the participants found this very upsetting. Some of the participants reported:

...I have had this disease for 14 years now...everything is getting worse. I get very frustrated. (M11) (Translated from Afrikaans)

But it's difficult and it's something that's going to get a whole lot worse. (M9)

Secondly, the participants reported that they found the anticipated life changes that would have to be made as a result of the progression of HD extremely challenging. Life changes can include having to move house, having to be cared for and being declared medically unfit to work. These challenges are described in the following excerpts:

...if things do deteriorate and you know I can't walk up the stairs anymore then we would have to move...Things like living on one level, we have got a lot of steps in this house... (F7)

What I would like and what is going to happen is probably different. Well I have just told my husband to please not put me in a nursing home...So for me it has been a big thing to come to terms with...I just think it is just, once again, not knowing. I don't

know if I will be able to stay at home and be cared for or I would have to go to nursing home and be cared for. (F5)

Whether or not I would be classified as medically boarded...it's going through the phases of the disease where you start to degenerate. (F2)

Lastly, a few participants were worried about their future because HD symptoms would only become worse. These participants are particularly worried about how they would manage to provide for their family and about the ensuing financial struggles. This is shown below:

I'm worried about the future. (M9)

I just struggled, I would go to bed, and would wake up and my mind would be racing, and I just think stress, and not being able to provide for my wife and that that sort of stuff and feeling a bit stressed...it's not a nice future. It's not something that anyone with HD wants to think about. (F2)

#### **4.3. Support/resources**

The next focus of the study was on supports and/or resources that participants felt that it helped them cope with HD. Supports and resources were identified on the micro-, meso-, macro- and chrono-system. The first level of Bronfenbrenner's Ecological System's Theory, the micro-system, consisted of almost half of the resources/supports and included: information, counselling, medication, coping, work, social support and the testing process. Thereafter, only one support was identified on the meso-system, which was the interaction between partner and family members and how that affected the participants. There were no supports or resources identified within the exo-system, meaning the next level was the macro-system. This level consisted of: cure, possible facilities, religion, disability grant, life

insurance and a medical aid. Lastly, there was only one resource within the chrono-system, namely, adaptation over time. These supports and resources will be discussed in-depth below.

#### **4.3.1. Micro-system**

**4.3.1.1. *knowledge about HD.*** Knowledge about HD was the first resource that helped participants cope with HD. This knowledge was gained through various sources, for example the internet, doctors and books about HD. Information served various purposes for these individuals, one being that they were informed of all the new research and updates that were happening in the field of HD, as one participant explained:

I mean, everyone I suppose is scouring the internet. I am signed up to about 6 different Huntington's companies on twitter, so obviously you get regular updates what might be happening around the world, so ya absolutely, helpful. (M1)

The following extract also conveys this sentiment:

...I think the research and the studies done, whether it's national or international...really do help to see, whether it's just a small inkling or whatever the story is... (F2)

Another way in which information was helpful was the fact that participants could assess situations better and make important informed decisions about their future, for example what happens to their body if they pass away. Two participants reported:

Firstly, it's knowledge that I can read and look at and maybe make decisions like would I maybe donate my body to medical science for it. (F2)

I like to know; and not knowing drives me insane. The more information I am able to get the better I can assess the situation and apply the right techniques to do what was necessary. (F5)

Information was also helpful to participants as it aided in their understanding of the disease as some had very little previous knowledge about HD. The participants' understanding of HD was aided by knowledge, in the sense that they did not only learn that there are others with HD, but also why people with HD behave in a certain way. All of these sentiments are conveyed below:

...I think with the proper education and knowledge it would change things for families with Huntington's. (F4)

I did a whole lot of research on the medication that she was on, on Huntington's itself...There was a lot of information available. I also bought the book: a family living with Huntington's. I read that book to try and understand and there were a lot of similarities in what she had gone through and what we went through as kids. So it was just reaching out to understand that there are others...it helped my understanding what was wrong with my mom, she used to do funny things...So we have a better understanding. I used to get upset with my mom a lot because every time she came to visit she would break something...So we started to understand all these things why she did the things she did. (F5)

Lastly, information can give you new ideas on how to help yourself or other loved ones with HD, as it could give you practical advice. This seemed to help, as one participant responded:



...if we had been able to say at that stage, Patrick, you have Huntington's and we must put little knobs on your shoes so that you can tie them and all those sort of things, I think that would have been so much better for everybody...it's having that knowledge is absolutely vital. (F7)

**4.3.1.3. counselling.** The second type of resource that participants found to be useful was going to a mental health practitioner, such as a psychologist, in order to help them to cope with HD. One participant described his psychologist as being helpful as he could talk about his problems:

...seeing a psychologist every 6 weeks or so. And she has been extremely helpful, just I mean, from a support point of view...Someone to talk to about all this kind of stuff, which has been absolutely amazing.' (M1) Another participant reiterated this when she reported: '...the psychiatrist,...has basically been pretty supportive...(F2)

Reasons why a participant expressed that her practitioner helped her was because she could disclose her problems but it would stay confidential, as well as having someone to talk to who would not pass judgement. These sentiments are expressed below:

Well I can speak to her in confidence. And I know that she will ask me how am I doing and I will be able to say listen this is what I'd like to do and she'll try and do some research and help me. It's basically knowing that there's someone that I can go to and speak to and that does not judge me. An independent person away from the emotion, away from the work situation... (F2)

Lastly, a participant mentioned how her counsellor is an important source of support. This is seen in the following quotation:

I go to counselling...She does the counselling, she is also familiar with Huntington's. So I go spend time once a month with her. It helps because it also keeps you on track with yourself so you don't lose the plot...She keeps me sane. She says to me, no you are fine. And I can always bring her into anywhere so that is nice, you know....I think counselling helps...Someone is on my side. (F8)

**4.3.1.3. medication.** Although there is no cure for HD, there are some medications that help with symptom relief. Medications that were taken included anti-depressants, tissue salts (cell salts or biochemical salts), sleeping tablets, and medication for the choreic movements. One of the participants mentioned that he was using tissue salts in order to help with his lack of concentration and it seemed to be working as he reported to be more focussed. The same participant also said that he was taking anti-depressants as a mood stabiliser. Two of the other participants were taking medication for sleep problems as they stated:

I went to speak to my house doctor and she offered antidepressants...but I did ask her for just sleeping tablets and she gave me 10 days' worth sleeping tablets. I said I'll be better if I can just sleep. If I can just think clearly I can work through my problems. (F5)

It does help...much more relaxed in bed at night I sleep better that's the melatonin that I take. (M9)

**4.3.1.4. coping.** Attitude also seemed to play a major role in helping participants cope with HD. One of the participants responded with:

It's going to be a little bad and then it's either way you going to take it. (F2)

Three more participants reported that a positive attitude was crucial to help them to cope with HD:

Attitude is everything. You have to have the right attitude you have to take it with a pinch of salt. (F4)

It's just, it's not all doom and gloom to get Huntington's...think it depends on how you see it and how you choose to live with it and that makes the difference I can wake up every morning and be angry and upset for having Huntington's, or I can wake up every morning and be seriously goal-driven to be the best mom and wife that I can be for as long as I can be. (F5)

Attitude plays a big role and when you are no longer able to convince yourself of these things (good things) then I think it starts to be difficult. (F8)

Almost half of the participants reported that acceptance of the disease was particularly important:

I think it would be stressful but I think maybe I've come to accept this is the situation... (F2)

Others also shared this attitude and reported:

If Huntington's happen, Huntington's happen. (F4)

I am ok with the disease. I have accepted it because I saw how my dad was. (F10)

(Translated from Afrikaans)

...the thought was well we all have to get old one day and if that is the way it was going to progress, the Huntington's route that is ok. (F7)

Another common view that was reported by five of the participants was the idea that, although they have HD and are well aware of the potential consequences of HD, they chose not to focus primarily on that. The participants reported that it was possible that they could pass away from something unrelated to HD, such as a car accident, long before HD became a reality. Some of their statements follow below:

...like I said I could live till I'm 80, it may never surface. Or it may surface when I'm 75 but then I died at 55 of something else, you see. (F4)

I could die from Huntington's but I could also be killed in a car crash for tomorrow. (F5)

Whether I live another 10 years, I don't know. I could live another 2 days, you know. I mean, I could get hit by a car and be gone by 2 days, or a week, or something, you know. (F8)

A third response was that participants seemed to make the most of everyday and they were not going to let life pass them by, as one participant said: '...I definitely make the most out of always like, but definitely make the most out of all the, all the opportunities now...' (F3)

Others affirmed this view and stated:

...and to live for now. So now is all I have. (F5)

I've lived my life to the full and Huntington's can go fuck itself. I am not going to sit around for anybody else. No ways. I am going to enjoy my life and that's it. That's where I have got to be because otherwise I'll spend every day in my closet, and there are days that I do. (F8)

So let's live each moment now that we can and enjoy things. (M9)

In addition to acceptance, passing away from an unrelated event and enjoying every day, comparing their situations to other negative events, such as other illnesses or financial crises seemed to help some of the participants to cope with HD.

Other families have cancer, other families have heart attacks, and other families have other problems, and it is all just an illness and it should all be treated as such. (F4)

I suppose the fact that there is everybody, everybody's lives are touched in some way with some issue or uhm, if Huntington's is something that we have to deal with in our family then I am ok with that...You look around the world and you just see what other people have to cope with and you just think oh my word. You know, poverty and wars and all those sort of things. I haven't got much...Huntington's...oh dear...I have got nothing to be sad about or angry about. None at all. (F7)

I don't have Diabetes, I don't have MS (Multiple Sclerosis), I don't have bronchitis, and I don't have asthma...I got Huntington's you know, great. Life hits whatever it will medically and so you just need to kind of go with whatever. (F8)

Lastly, what also seemed to help participants was that they approached HD on a day by day basis and tried not to think too far into the future, but rather concentrated on the here and now.

...take one day at a time you know. (F2)

You take a month at a time, you take a year at a time; you see where you are. (F8)

I think we just learnt not to think of the future we live each day as it comes and handle that. (M9)

For me it's also a day by day thing...It's a day by day thing. You just have to take it now day by day. (M12) (Translated from Afrikaans)

**4.3.1.4. employment.** Although it was previously mentioned that some participants found certain aspects of employment to be a challenge, others stated that it was in fact a resource for them to help them cope with HD. One of the reasons why it was considered to be supportive was because it kept participants busy and if they were busy they were less likely to think about HD. This is conveyed in the excerpts below:

There isn't really time to talk about anything else during the day. Definitely...yes, definitely helpful. (M12) (Translated from Afrikaans)

You can't think of anything else you know...but I do find that when I'm...not as busy...then my mind does wander...so I definitely prefer to be kept busy ...I like the fact that I'm busy at work. I love my job and I don't even think about it really...and that helps. (F3)

It (work) keeps me busy and motivated. (F4)

In addition to keeping participants busy, colleagues and bosses were also described as an important source of support for some. This was mainly because participants were fortunate enough to receive special treatment such as reduced work load or permission to leave early for the predictive testing process. Colleagues were also reported to be supportive as they offered emotional support to participants. These sentiments are described in the next extracts:

At the moment I've actually got a really nice manager I'm working for. And the work load is much less. But previously I had a lot of workload I had to attend to. Say last year and I told the manager I wasn't coping at all. (F2)

...people at work all know about it. So they also are very helpful. Like when I went through the testing process. I had to leave you know early a couple of days and all of that and they were always very helpful about that, so *yes* [ya]. (F3)

They support me a lot and I have an amazing boss at the work and at work I have just as an amazing support network. Everyone is there and the acceptance of me as a person and they know what is going to happen, what is going to happen to me, what lies ahead of me and they support me. When I arrive at work and I look annoyed, you know, then they will ask, 'what is the matter?' Then you know something is wrong. So we can talk about it. It's really an amazing experience at work. (M12) (Translated from Afrikaans)

**4.3.1.6. social support.** The next type of resource that was identified as being part of the micro-system was social support. Spouses, families, friends and the support group all formed part of participants' social support. The first type of support was the participants' spouses. What seemed to help participants was the fact that their spouses were informed about HD, as this prepared them for what is to come in the future. This is described below:

Any other queries or questions, about the process, about me, about the future, about genetics, at least, after the 6 months, before I got the test, the results, you know she was incredibly informed. (M1)

There is nothing he doesn't know, so my suggestion to any person who has got Huntington's, rather share the information and knowledge. (F4)

I know so I am prepared and my husband knows and so...we as a family unit understand what is going on and how it works. So it is not a discussion, it's not something I am hiding. (F5)

It was also noted that participants appreciated that their spouses did not leave them when they found out that they tested positive for HD. As one participant stated:

...he also never walked out on me, he has stuck around for the last 10 years...Give him major credit for that because he could have walked out, quite easily and decided it's too much for him.' (F8)

There were also others who shared this sentiment:

He has been an unconditional rock, from day one he has said don't worry, I am here, we will deal with it whatever comes across our road, I will not be gone; I will look after you. And every time I have worried about it, he has always reassured me to say to me, don't worry. (F5)

At that my stage of life, with a stable relationship...I wasn't worried that it was going to affect my relationship with my husband so...but one can't say that for everybody at all. (F7)

Lastly, the participants reported that having their spouses treat them as still normal seemed to help them cope with HD. This is expressed in the following two excerpts:

My husband said it's the same, now that you have been found to be positive, that is fine. It doesn't change anything from his point of view, that is just the way it is...Not to make an issue of it. (F7)

A wonderful wife...she treats me like I'm normal and not like I'm sick. (M9)

Friends were also mentioned as being a support for some, although this was not reported by many. Only two participants mentioned that their friends were viewed as an important resource for them. One of the participants said:



I mean all my friends have just been also very supportive and just encouraging and just getting involved where they can...(F3)

Another participant reported:

We are part of an amazing support network, not only us as a family, but extended family and friends... (M12)

Next were the participants' families who were described as being an important resource for them. As one participant mentioned:

I don't know if the word independent is incorrect, but I draw strength from my family and I place them very high on my priority list, so as long as they are there I can tackle anything. (F5)

Although HD was previously described as a 'monster', it still seemed to bring families together, as a participant described:

You know, this disease, as horrible as it is, it has really brought our family together so much. We have been very very lucky. (M1)

Other participants had similar experiences within their families and reported:

*Yes* [Ya], as you know my father passed away about eight years ago of Huntington's disease, so it's just my mom that's around and *yes* [ya] I think that's obviously had a reason for our closeness. (F2)

...but then we decided that as a family, this thing will either tear us apart or bring us together and it really did bring us as a family closer. (M12) (Translated from Afrikaans)

During the testing process also, family seemed to offer a lot of support to the participants. In order to be tested for the HD gene, you need someone to go through all of the steps with you, and participants who were not married reverted to their families for this support. Two of the participants asked their mothers to support them:

...basically my mom's been with me right from the beginning to the actual, the actual result...So I think she's really helped me a lot. (F2)

You're not allowed to do it by yourself. So like my mom came with me. You have to have like a buddy. (F3)

A number of the participants also explained how watching family members with HD made acceptance of HD easier. It encouraged them to see how their family members coped with HD. Family members with HD also contributed to prepare the participants for their future because they had a good idea what the progression of the disease would typically be like. These sentiments are expressed below:

...my dad...he was the carrier...he was just such a person that got on with everybody and I thought, if that is what my old age is going to be like, then I can manage that. (F7)

I am ok with the disease. I have accepted it because I saw how my dad was. (F10)  
(Translated from Afrikaans)

Uhm, it's been a good eye opener for me. You get the raw whatever, up straight. So you are not beating around the bush, which is nice. You know exactly what you are dealing with. (F8)

I think having seen my side of the family; we know what to anticipate...we at least know what's coming, you know...There are no surprises... (M9)

Talking within the family about HD was also considered to be an important factor in helping participants cope with HD. Some participants mentioned how they could talk freely among family members about HD and that seemed to help them. One of the participants reported:

We talk about this the whole time...we were always in communication with all of our aunts and uncles...you know and about latest outcomes about Huntington's. From a support side of things, I have been very very lucky. (M1)

It was also valuable when other family members talked about HD openly, so as to encourage talking throughout the family. This is expressed below:

I think because she's always been very open about talking about it. She's never hidden or tried to hide anything so I think that's been a big asset because you sort of do feel you've got the cousins of course that's got stuff in common and you can talk about it. (F6)

The last reason why family was considered to be a support for individuals living with HD was because participants whose HD was more progressed needed more physical help, such as with preparing of meals. Some family members took over this task, as stated by one of the participants:

Yes my mom looks after me. And my daughter makes food. It helps. Otherwise I break things. (F10) (Translated from Afrikaans)

The support group, Huntington's Association of South Africa, also formed an integral part of the social support that was described by the participants. The support group was mentioned as being helpful because there were other people with HD, professionals would come to present talks, and they could share their experiences within the group, which participants felt was encouraging. The first reason why it was considered to be a support was that individuals felt that the group encouraged them and gave them hope, as one participant reported:

There's first of all HASA, Huntington's Association of South Africa...It just...it makes me feel like yes I can do it I can sort of cope and take one day at a time you know. (F2)

Another participant shared this view:

...before I was kind of involved in the support group, I looked at it as a death sentence you know but like not at all anymore. (F3)

Another important aspect of the support group was that there were others with HD as well. This was important as people felt less intimidated by HD when they were in contact with other people who had HD, and they also felt that they could relate to the others at the support group meeting. These sentiments are conveyed by the following excerpts:

And especially if you deal with these types of people or if you're in contact with people on a daily basis, you know, you sort of becomes like, you're not scared of it anymore. (F2)

...the fact that they get to meet other people who have also heard about this condition because everybody at the very first support group...was just like in shock to be with other people that knew what they were talking about...it was so nice of them to

finally, for me as well was like I had people to relate to.....with the support group you don't have to explain...to people what it is that his got because they just know. (F3)

Some support group meetings also have professionals with expertise in the field of HD such as financial and medical aid advisors, genetic counsellors and genetic researchers to speak to the members. This seemed to help the participants because important information is discussed at these meetings as these professionals share their expertise, as well as encouraging participants with new research developments. This is seen in the following statements:

I'm just sorry because apparently they had one guy in about finances and medical aid. Those two would have been very critical for me...it helps, there are a lot of loop holes and you can get yourself into big trouble if you don't know, you know, you need to know what you are on about. (F8)

I think the fact that there are professionals...like, Copperberg (pseudonym), ya then you know you are able to listen to those people...they give you hope and think well, they seem so dedicated to the task that you just think wow that is fantastic. I have got this extraordinary thing and there are people out there who really care about trying to help people in this situation and trying to do research to get medication that can also help. (F7)

The last important function that support groups fulfilled was that it encouraged talking among the members about HD. This was useful as information and practical advice were shared or just simply sharing experiences within the group. The following statements convey this sentiment:

And the fact that I love it when somebody in the support group will say something like *yes* [ya] like the person's getting a bit...you know the person always wants to eat and I don't know what to do and then somebody else in the support group will say just give him, just keep Pro-Nutro you know...When someone like is like really struggling with someone, with something... (F3)

...these discussion groups too you know. It's important that people are able to sort of go and feel what or say what they're feeling and really get it out there and *yes* [ya] no there's certainly a lot more....I think just being able to talk freely to people who either have had it or have experienced it. (F6)

**4.3.1.7. testing process.** The last resource that was identified as part of the micro-system was the testing process, although previously stated as being a challenge for some. The testing process consisted of the medical staff that was involved, external support and the results. First of all, the medical staff that was involved in the testing process was described in a positive manner by several of the participants. One of the participants reported: '...we were very lucky. We had Maggie Smith (pseudonym)...' (M1), and this sentiment was shared by others below:

...I mean she's remarkable she really is...sister Claire (pseudonym) was there the whole time... it was good to get to know those people... (F6)

...they (staff) were excellent. (M9)

One of the reasons why the staff was viewed positively and as helpful was because all the information, which included a lot of details about the condition itself, was conveyed to the participants in an understandable manner. This is evident from the quotes below:

But obviously, its, informative and that is the case. (M1)

The genetic counsellor is that she explained exactly what the gene was and the biological form....There was a lot of things that I did not understand that she tried to break it down as simple as possible...counselling sessions on what is the disease, what will happen, what impact it will be on you and also for your family too. (F2)

The staff also did their best to prepare the participants for the test results and this was appreciated by the participants. The testing process was described as being comprehensive and meticulous as one participant stated: 'It was thorough. It was completely thorough.' (F4) This was because of all the different specialists, such as psychologists and neurologists, who were involved during the testing process before the participants received their results. This made the participants feel more prepared for their results and was stated as:

They were very supportive and you know they had made us see a psychologist and everything before they gave us the results. (M9)

But not only that, it was the neuropsychology sessions and all those sorts of things...it was quite amazing. (F7)

You have a session and the person you went with, just to make sure that you can, you can handle the result. You know that you're gonna be okay. That you can cope. That your life will kind of go on, or like at least as good as it can... (F3)

Participants were also prepared for the test results by not rushing the testing process; the results are often withheld until the individuals are ready to cope with the feedback. This was the particular case with one participant who was denied her results because the professionals believed that she would be unable to cope with the outcomes of the tests; she said: 'They said to me I wasn't ready for the result.' (F8) The reason for this purposeful delay was to ensure that the participants were thoroughly prepared, by making sure that firstly, they

wanted to receive the results, secondly, that they were doing it for the right reasons, and lastly, by giving them ample opportunities to opt out of the process.[C1] [C2] These sentiments are described below:

...but it's not that it takes that long. It's just that you need to go through a whole lot of, they wanna make sure that you're like ready for the results...So it's quite drawn out but it's you have a lot of time to make sure that it's what you wanna do. You have a lot of time...pull out if you don't want to. (F3)

They wanted to make sure that you were not doing it for anyone else but only for yourself. (F4)

They did blood tests and then they gave us the option at anytime not to get the results but yes [ya] we were given fairly thorough...what was it once a month that we had to go? (F6)

Participants were also allowed to have someone go through the entire testing process with them, as the medical staff felt that this would be additional support for them. This really seemed to help the participants cope better with the tests and results because the process was long and worrying. This is described in the following excerpts:

...she went through the entire testing process with me, which is obviously a huge thing you know. So as you know, the process is 6 months, and she went with me to every single specialist. (M1)

Also I was allowed two people to sit in with me for support, as it can get quite emotional or stressful and so I asked if my mom to sit in with me and then the person I was involved with. (F2)



You're not allowed to do it by yourself. So like my mom came with me. You have to have like a buddy...I think it's a good setup that you need to do it with someone. (F3)

In retrospect, all the participants reported that they would still want the results, even though they knew now what the outcome of the test was.

Yes, without a doubt...And I think it was one of the best things that I could have done. (F2)

I would definitely still have the test...I thought for myself I needed to know yes. (F4)

I think I would prefer to know. (F6)

According to the participants, the test results changed their perspectives on life. They reportedly appreciated life more and this contributed to them living for the moment and enjoying every opportunity they had. This sentiment is described in the quotations hereafter:

If I think, anything else, you just appreciate everything more, especially now, and that we have a baby girl, so we are just enjoying every single day as much as possible. (M1)

...I managed to at the age of 30 when I got my results, you know, to enjoy life, see things differently and change the things I want to change and do the things I want to do so for me it has had a positive result...and to live for now. So now is all I have. (F5)

You don't understand, I couldn't see the world before, now I see the world. (M12)  
(Translated from Afrikaans)

The test results also made it easier to plan for their future as the participants knew that they had to make provision in case HD did develop or progress further. Planning involved reconsidering what they wanted to do with their lives, living arrangements as well as acquiring policies, such as life insurance, and testaments. These are all conveyed by the following excerpts:

I would rather be positive and plan the rest of my life. (M1)

The test has meant for me, basically what is basically analysis of really what do I want in my life. (F2)

Like I said, practical stuff in life. Insurance policies and your home, if I knew I tested positive then I wouldn't buy a double storey house because that would be difficult to get around and yes [ya], practical things, which makes a difference to me. (F4)

I think knowing the results, you are able to put things in place that need to be in place, like a life insurance policy and to be able to make the decisions of how you are going to be looked after when you can't look after yourself anymore. (F5)

Lastly, several of the participants reported that knowing that the outcome of the test result was positive was better for them than not knowing if they have HD or not. Before they knew about the outcome of the test results, many of the participants were in fact anxious about their unknown status, as can be seen by the participant's response:

I always wanted to know, it always bothered me...I used to have sleepless nights....it always bothered me before the testing; not knowing. Fear of the unknown... (F4)

This anxiety was relieved once they found out that they did in fact have HD. Therefore, test results were considered as a resource for the participants to cope with HD, and can be seen in

the following statements:

And the fact that I don't want to live a life of ignorance and also the fact that it gave me an opportunity for me to say...there's going to be a result. (F2)

No, I wanted to know...because you see now I know. I am glad I know; I am happy that I know. It calmed me down... (F4)

...it was ok, because I knew, I knew. What makes it easier now, the blood tests gives you a definitive answer, which makes it very easy...It gives you a straight answer otherwise you are dealing with an unknown. I don't want to deal with an unknown. That is a very big thing to deal with. And you need a straight answer you know. (F8)

I prefer that I know. The fear of not knowing is a lot worse than the fear of knowing because now I know what awaits me, because if I didn't know, then I'll always be wondering, but now I know... (M12) (Translated from Afrikaans)

#### **4.3.2. Meso-system.**

The next system where supports were identified was the meso-system, consisting of interactions between the individual's different micro-systems. There was only one resource identified as being a part of the meso-system, namely partners and family members with HD. This resource consists of the interaction between the participants' partners and their family members with HD and the positive consequences thereof.

**4.3.2.1. partners and family members with HD.** This was considered to be part of the meso-system as it was clear that there was some interaction between the participants' partners and their family members, both included in the micro-system as discussed earlier. It was considered a support to have participants' partners or spouses exposed to the family members

with HD, as there were in fact some positive spin offs. Those participants who were in a relationship felt that having their partners see their family members with HD helped prepare them for their possible future as their partners could see what the progression of HD would look like. This meant that their partners would not have any illusions about HD. This is conveyed in the following sentiments:

...she literally saw him at his worst. So lucky enough for me, that obviously for me she had known and had no inhibitions about what might happen to me one day, so I was very very lucky that she had seen the worst case scenario. You know. So ya, I think in that respect I have it quite lucky that my wife possibly knows what to expect in the future. (M1)

Yes, he has been around of 10 years of this. He was also in the house watching my mother go down. He has known for 10 years that I have Huntington's. He was there when I got the diagnosis. (F8)

Others felt that having their partners see family members with HD was helpful as they could not hide the fact that they have HD in the relationship.. As one participants stated:

So he's very much exposed to it (family members) and you know so I can't just pretend it doesn't exist. Which I think is good. I can't just pretend. (F3)

#### **4.3.3. Macro-system**

There were no resources or supports identified in the exo-system. This was because the exo-system consists of links between the micro-system but where the individual is not actively involved, for example the participant's partner and the partner's workplace. As the study focussed on the individual's experiences, there were no resources identified on this level. Therefore the next level where resources and supports were identified is the macro-

system. The macro-system consists of the individual's culture and values, as well as encompassing the laws and policy making, which affects the individual. There were several resources identified as being helpful in order to help the participants to cope with HD on this level, which includes: medical aid, life insurance, a cure, possible HD facilities, religion and disability grants. These resources are all discussed in the following section.

**4.3.3.1. medical aid.** The first resource that was found to be part of the macro-system was a medical aid to help with the treatment and medical expenses related to HD. As was evident in the previous section on challenges, most participants viewed medical aid as a challenge, firstly because it is expensive and secondly because medical aids generally cover only limited medication that is needed for the treatment of HD. However, there were two participants who found that their medical aids were helpful because they were flexible, in the sense that you did not have to be on the most expensive plan but could change once you started showing symptoms and required more benefits. This is stated below:

...the thing about Discovery is you can upgrade or downgrade once a year, so...So I can be in the bottom one and then feel like okay I need to up my medical aid. (F3)

Participants also indicated that having a medical aid provide them with peace of mind:

So the fact that Discovery gives us anything I'm happy with. (F3)

...should I get at a point where I need medication or whatever, I have medical. (F4)

**4.3.3.2. life insurance.** Another resource that was previously reported as a challenge is getting life insurance for when participants cannot work anymore or when they pass away. This type of insurance generally includes disability insurance for when they can't work anymore. Participants explained that if life and/or disability insurance were arranged before

they went for predictive testing then there were no problems with getting the cover. This is explained below:

I always knew that I need to get that done before, my financial advisor said to me if you decide to get tested just know that you need to have all your cover in place first, so I was always very aware of that. (F3)

We did all that beforehand. So we handled it, especially disability and stuff we had all that...just before everything came out on my side. (M9)

The reason why insurance policies were deemed important was because it took care of the financial concerns that participants had for the future when they could no longer work. This money would help their families so that there would not be a financial deficit in the families' monthly income. This can be seen in the following quotations:

So yes [ya] like put a decent amount of money away every month...I don't wanna have to, like if I'm sick the last thing I want my husband to worry about is money...And like I said I know it costs a lot you now but I know it's going to be worth it... (F3)

It allows you to know that if I can't work anymore then there is a policy that will pay out so that John (pseudonym) doesn't have a financial deficit in our monthly responsibilities. If I pass away, then there is something that will pay out so that he doesn't have to worry. (F5)

...I do have like a financial life insurance premium through the company every month...And I also have my own personal disability insurance which is paid out. So honestly what the premiums do financially, it should be equal like a monthly amount. (M9)

**4.3.3.1. cure.** Another support that participants mentioned that helped them to cope with HD was their hope for a cure for HD in the near future, as one participant reported: '...the hope that something would come up medically.' (M9) With the new medical advancements, such as human trials to stop the HD gene, participants were more hopeful towards their future as they realised that by the time they started to show signs of HD that there will be more therapies available to help. These sentiments are described by two more participants below:

...there's so much positive stuff happening and you know they're doing human trials...it is just around the corner...so yes [ya] just difficult not to be positive about it...I just feel like life can go on...and I can live a normal life and I know I'm not starting to show signs, so hopefully let's say if I start showing three or four years or five years time, there'll be something there you know...is extremely encouraging. (F3)

Although we live obviously in hope that the clinical trials that they're...going to start next year will produce some medicine that will retard it...Whether it will reverse it, we're not sure but so mean that's the sort of hope that one talk about you know? (F6)

The same hope for a cure was also expressed for the participants' children. It seemed to be comforting to them that there will hopefully be a cure for the next generation, and parents were positive about their children's future as there are more developments regarding the treatment of HD. These statements are seen in the following quotations:

...by the time our children possibly show Huntington's symptoms, will be in 20 to 30 years, we would like to think there is a cure by then. (M1)

...but now that there's some stuff happening, I'm not even, been worried about that because by the time my baby is like 30, if there's human trials in like three years time,

by the time my baby's going to...the earliest time the child might start showing is 30, do you know what I mean? Then there's going to definitely be a cure by then. (F3)

And I certainly think for the kids you know thinking about having children of their own, I think at least they know there's an option possibly that you know they can have that little test...That little Petri baby...because now there's more positive news about that hopefully. (F6)

As a result of all the knowledge and medical advancements that is being generated in the line of HD, participants seemed to be positive and optimistic about their future, as one exclaimed:

Normal with lots of babies!...There is so much happening to create awareness and funds internationally, so just trying to remain as optimistic as possible! (F3)

Other responses included:

Yes, I think because now there's more positive news...there's so many companies doing research and countries doing research on it...there is light at the end of the tunnel whereas before there wasn't much. (F6)

Bright and colourful. (M12)

**4.3.3.4. possible HD facilities.** In addition to medical and life insurance and a cure, participants also found comfort in knowing that there will be a few beds available for people with HD in the near future. There will be 32 beds allocated for individuals with HD at the Valkenberg Psychiatric hospital in Cape Town in the Western Cape province. Half of the beds will be for respite patients and the other will be for long-term stay. Knowing about these beds and that there would be specialised care for people with HD came as a major relief to



some, as one participant reported: '...but I just felt like a weight's been lifted off my shoulders.'(F3) Others shared this feeling of relief, in addition to being elated and feeling calmer about their future in case they would need to be moved into a special care unit, as they know there would be specialised doctors to help them. These statements are described below:

I mean 5 beds, that's a break through. (F4)

If there is a place...I actually feel so at peace about that because it is such a big step... if you have a place at Valkenberg, you have got doctors on tap and nurses and things who are just around looking after people with Huntington's, specifically I just think it is going to be so much better. (F7)

Facilities were also considered a resource when the participants themselves had to look after their family members with HD. It was helpful when participants could take their family members to a specialised home so that they did not have to look after them constantly and could take a well needed break. This is described below by one of the participants as she speaks about caring for her mother:

Putting her into a place where she had 24 hour care, where someone is looking after her and to relieve me. (F8)

**4.3.3.6. religion.** Only three of the participants reported that religion was an important resource to help them to cope with HD. However, it is evident from their responses that this was an important resource.

Yes, Christianity definitely. You can't do these things without the Lord and place it in the hands of the Lord. (F4)

I can give it all to Jesus; I don't have to hang onto it. (F8)

Religion was also used to help them to keep their HD in perspective. It was stated that their condition, in relationship to God, was insignificantly small and one should not allow it to get bigger than it actually is. This sentiment is described below:

What I have also done is that I have put a pencil dot on the wall, like this, ok, that is my Huntington's. The rest of the wall is God. God has got my Huntington's, the rest of the wall and everyone. As long as I keep everything in perspective, there is my dot, which is my Huntington's, so I don't allow the issue to get bigger. (F8)

With keeping HD in perspective, participants were also comforted by the fact that they knew their body was a temporary physical entity, and that one day they would go to heaven and would be relieved of their body with problems. This is conveyed in the following sentiment:

You get a body, you get health issues with the body and you get your own personal what you get handed out, but one day I will go to heaven. One day I will be part of God and I won't have this stupid body that is flawed and has got illness and has got whatever. So when you look at it that way, it kind of shifts a whole perspective on your life. (F8)

Lastly, it was also stated that participants' religion gave them motivation every day, allowed them to make peace with their condition and contributed to giving them a purpose in life. These sentiments are conveyed by the following excerpt:

People's hope and peace come from others and earthly possessions. However, as Christians, hope and peace come from God; He gives us peace and love, and He is that. My power and strength comes from Him. That is what drives me to live day for

day. Without Him, I am just another soul that walks spins around in this whirlwind of a life that we live in. (M12) (Translated from Afrikaans)

**4.3.3.5. grant.** The last resource that emerged on the macro-system was a disability grant from the government. As previously mentioned, some participants lost their work as a result of HD. However, HD is now recognised as a disability and individuals are allowed to claim disability grants from the government, as stated by a participant:

I mean, look at the disability grant. Until recently, people with Huntington's didn't even get a disability grant you know. It's very recent thing also. (F4)

Two of the participants made use of this benefit:

Only from Groote Schuur, they gave me a grant. (F10) (Translated from Afrikaans)

I am going to get one now. (M11) (Translated from Afrikaans)

#### **4.3.4. Chrono-system.**

The final level of Bronfenbrenner's ecological model is the chrono-system. This level consists of the changes or consistencies that happen over time in the individual's life span. There was only one resource that emerged on this level, and consists of the participants' abilities to adapt to the changes, posed by HD, over time.

**4.3.4.1. adaptation over time.** Participants explained that although HD is a progressive disease, the changes that they would have to face would happen slowly. This was important to them as they felt assured that they would have adequate time to modify their lifestyle and to bring about the necessary changes they would need to make. The following quotations convey this sentiment:

Uhm, I feel that if there is you know, if things are going to get worse, they are not going to get worse in a hurry and I think we will all be able to adapt accordingly...to put things in place. (F6)

I think we've just had so long to get use to it that you know and...we just take each day and deal with each little quirk as it comes. (M9)

Some of these changes would have had to been made because of their age, even if HD never developed, such as having to move houses, in order to be on one level as to avoid stairs. It seemed that participants were more positive about changes if these changes were related to ageing and not to HD. One of the participants stated:

...if things do deteriorate and you know I can't walk up the stairs anymore then we would have to move but that would have had to happen anyway. We are already 63 and we are getting to that stage we need to help each other along and that sort of thing. (F7)

## **Chapter 5**

### **Discussion**

This chapter, which is also the final chapter, will consist of a discussion of the results that emerged in this study and integrate it with existing literature. The strengths as well as limitations of this study will also be discussed. Recommendations for future studies regarding this type of study will additionally be made. Concluding remarks will follow.

#### **5.1. Introduction**

Bronfenbrenner's Ecological System's Theory, or Human Ecology Theory, will serve as the theoretical framework to facilitate the interpretation of the results as well as to contextualise the discussion.

Themes emerged on all the levels of Bronfenbrenner's (1979) Ecological System's Theory, apart from the exo-system. The majority of both challenges and supports were identified as belonging to the micro-system, which included direct and intimate interactions of the participants. This was expected as the focus of the study was on the individual with HD and their experiences concerning their condition. Although not the focus of this study, there was one challenge and one support identified that belonged to the meso-system. These themes consisted of the relationships between the individual with HD's spouses and their families. There were no challenges or supports identified on the exo-system and this could be because the focus of this study was on the individual who was living with HD and not on those around them. Challenges and resources or supports were also identified on the macro- and chrono-system, which included policy making and changes over time respectively.

## 5.2. Challenges

### 5.2.1 Micro-system.

The micro-system, as previously mentioned, consists of intimate and direct social interactions that occur between an individual and others around him/her (Bronfenbrenner, 1979). Challenges were the first main theme that emerged from participants' responses and consisted of eight subthemes, which included: triad of symptoms, sleep problems, testing process, relationships, children, it's a monster, employment and lastly, social support. It was evident from the data analysis that individuals with HD experience several challenges due to their condition and that HD is a devastating genetic, neurological condition.

**5.2.1.1. triad of symptoms.** The interviews naturally started with participants' descriptions of the variety of symptoms that relate to HD. So, the first challenge that emerged on the micro-system related to the triad of symptoms that individuals with HD experience. The triad of symptoms include motor disturbances, cognitive dysfunction and behavioural problems and is widely recognised in literature about HD (Gudesblatt & Tarsy, 2011; Ha & Fung, 2012; Haddad & Cummings, 1997; Roos, 2010). The majority of the symptoms reported by the participants were concerned with motor disturbances, as some form of motor difficulty was mentioned by half of the participants. Motor disturbances reported by participants in this study included uncontrollable movements, balance problems, which affected walking, resulted in dropping of objects, and speech and swallowing difficulties. All of these symptoms have been studied extensively, and are in accordance with the literature (Dalton et al., 2013; Gudesblatt & Tarsy, 2011; Heemskerk & Roos, 2011; Vogel, Shirbin, Churchyard, & Stout, 2012) Motor disturbances, in addition to a family history of HD, are one of the determinants of making an official diagnosis of HD (Gudesblatt & Tarsy, 2011; Roos, 2010). A prominent motor disturbance according to the literature is eye movement

abnormalities (Blekher et al., 2006; Dursun, Burke, Andrews, Mlynik-Szmid & Reveley, 2000; Patel, Jankovic, Hood, Jeter & Sereno, 2012); however, none of the participants reported this to be a challenge. This may be because many of the participants in this study were in the prodromal phase (before disease onset) of HD and did not display many HD motor symptoms yet. In addition, it may also suggest that eye movement abnormalities are perceived by participants as less severe than other motor abnormalities, for example, chorea.

Cognitive symptoms were the next group of symptoms that were reported by the participants and also form part of the triad of HD symptoms. It should be kept in mind that these symptoms related to cognitive disturbances that were perceived by the participants because no formal cognitive tests were administered. It is important to identify cognitive signs early because the onset of cognitive symptoms in individuals with HD may appear 15 years prior to motor disturbances (Duff et al., 2010). Only two cognitive impairments, namely a lack of concentration and memory problems, seemed to be prominent in the experiences of the participants in this study. This may be because participants were only aware of these two problems, as it clearly impacts on their daily lives. However, it does not necessarily mean that other cognitive signs such as problems with emotion recognition or visuo-spatial problems were not present, it may just be that the participants were not aware of them. Decreased concentration seemed to hinder progress at individuals' place of employment as tasks were completed with great difficulty. Concentration difficulties also affected their driving and seemed to worsen as the day progressed. Previous research suggests that attention impairments may underlie many of the other cognitive problems that are associated with HD, such as emotion recognition, language difficulties and visuo-spatial problems (Sprengelmeyer, Lange, & Homberg, 1995). A study conducted by Zakzanis (1998) suggests that concentration and attention was consistently slower in individuals with

HD, rather than showing a deficit in attention. Regardless of the mechanisms that underlie attentional problems, it has been well documented that attention and concentration is a major challenge for individuals with HD (Aron, Sahakian, & Robbins, 2003; Snowden, Craufurd, Griffiths, Thompson & Neary, 2001; Thompson et al., 2010). According to existing literature on attentional slowing in individuals with HD, the comment by participants that 'concentration worsens as the day progresses' has been unfounded to my knowledge. This might be because these studies administered tests over a relative short period of time, instead of simulating a working day, e.g. completing tests early in the morning and then late afternoon.

The second cognitive impairment reported by participants was that their memory seemed to be affected by HD. One participant reported for example that s/he often forgets his/her wallet at home when going to the shops. It is clear from the literature that memory problems, more specifically implicit- (Maki, Bylsma & Brandt, 2000), procedural - (Butters, Wolfe, Martone, Granholm, & Cermak, 1985) and episodic memory deficits (Montoya et al., 2006) have also been identified as a major cognitive component of HD. Memory involves three steps, which are acquiring information, storing it and then lastly, retrieving it (Bourne et al., 2006). Ring and Serra-Mestres (2002) state that it is the third stage, retrieval of information, which individuals with HD generally struggle with the most. A decline in cognitive function has been described as the most disabling component of HD (Rothlind, Bylsma, Peyser, Folstein, & Brandt, 1993). This was emphasised by one of the participants in this study who reported that she was scared that she would not be able to learn new information or help her family in emergency situations when she started to present with cognitive symptoms.



The last of the triad of symptoms were psychological or behavioural problems. Feelings of depression were the most frequently reported challenge, not only in the literature (e.g. Roos, 2010) but also by most of the participants in this study. A low mood was mostly experienced shortly after receiving their genetic results. Also in accordance with other studies (Boot & Chaudhuri, 2012; Haddad & Cummings, 1997), depression often preceded motor symptoms. Only one participant reported a personality change in that she experienced a lot of anger for which she was taking medication. Although high frequencies of suicidal thoughts in individuals with HD have been previously reported (insert references), it did not seem that any of the participants in the current study could be classified as being suicidal. However, it was not explicitly asked whether participants had experienced suicidal thoughts, which may be why none of the participants reported this. It is also possible that because only one interview was scheduled with each participant that they chose not to report this as a challenge, because of the personal and sensitive nature of the information. Suicidal ideation is a very personal matter, which may require a relationship that is more long-term in nature, in order to build trust and rapport. Other psychological symptoms that have been cited in studies but which were unfounded in this study include: lack of interest in life, increased irritability, impulsivity, hallucinations, mania and delusions (Gudesblatt & Tarsy, 2011; Haddad & Cummings, 1997; Paulsen et al., 2011; Quarrel, 2008). Again, this may be because the participants were simply unaware of these symptoms or simply chose not to report them. Participants were assured that they only had to report what they felt comfortable to share. Self-esteem issues, due to involuntary movements caused by HD, were also reported in this study, which, according to Roos (2010) and Quarrell (2008) are common among individuals with HD.

**5.2.1.2. *sleep problems.*** Participants also experienced several sleeping problems; some of the participants fell asleep quickly, some struggled to fall asleep and wake up, and some of them experienced a lot of movement either before falling asleep or during sleep. Aziz et al. (2010) studied the nature of sleep disturbances in individuals with diagnosed HD and found that night-time sleep impairments included a significant delayed sleep onset time, higher use of sleep medication, longer sleep duration and daytime dysfunction (Aziz, et al., 2010). This study also found that individuals with HD experienced a significant delay in the usual wake-up time (Aziz, et al., 2010). The delayed wake-up time was found to be associated with a decreased cognitive score and functional capacity and an increase in depressive symptoms (Aziz, et al., 2010). Another study that looked at sleep problems in individuals with HD was performed by Arnulf et al. (2008). Compared to the healthy control group, individuals with HD presented with lower sleep efficiency (the number of minutes of sleep, which is divided by the number of minutes an individual is in bed), as well as a longer wakefulness duration after onset of sleep, which indicates problems with maintaining sleep (Arnulf et al., 2008). It was also observed that individuals with HD experience more periodic leg movements compared to the control group, which causes more arousal during sleep (Arnulf et al., 2008). Although the participants who reported to fall asleep quickly described it as being positive, it could be a sign of sleep deprivation when sleep onset latency is shorter than five minutes (Dement & Vaughan, 1999). These types of sleep problems that are experienced by individuals with HD can potentially affect their quality of life, be a potential risk factor for injury, as well as be a risk factor for depression (Arnulf et al., 2008; Aziz, et al., 2010). It is evident from the results of this study as well as the literature that the triad of symptoms that individuals with HD present with can have an enormous impact on their daily lives.

**5.2.1.3. testing process.** The next challenge that emerged from the results related to the testing process of getting their HD results. There are many reasons why individuals with HD decide to go for predictive testing. A study conducted by Futter et al. (2009) that evaluated predictive testing in the Western Cape, South Africa, found that the main reasons for testing for HD were: to reduce anxiety and uncertainty (62.9%), to plan their future (59.2%), having responsibilities (33.3%), informing children of their status (25.9%), guilt about passing on the HD gene (25.9%), informing their children of their personal status (22.2%) and family planning (18.55%). All of the participants in this study reported a combination of these reasons as why they chose to go for predictive testing. There seems to be many challenges related to the predictive testing process. Some of these challenges that have been reported by the participants of this study included: there were too many health professionals involved during the testing process, some tests seemed unnecessary and were not properly explained, some aspects of the process were not thorough enough, especially follow up sessions, it was a lengthy process, some participants had to travel very far, and most of the participants experienced some intense adverse emotion as part of their reaction to their results. Unfortunately, some participants had to travel far distances as there are only 4 testing centres in the whole of South Africa (Krause & Greenberg, 2008). The participants who were shocked by their results stated that they did not anticipate the positive HD results. This has been shown to happen when individuals go through the testing process and secretly believe that they do not have the HD gene, and then are in a state of disbelief when they receive a positive result (Whalin et al., 1997). Other emotions that participants reported in reaction to their positive results were anger, guilt, and disappointment. All of these emotions have been reported in other studies (Tibben, 2007 van 't Spijker & ten Kroode, 1997).

**5.2.1.4. relationships.** In addition to symptoms, sleep problems and challenges with the testing process, participants also reported that their results had an effect on their relationships with their partners, whether it was an existing or future relationship. Increased levels of distress, often due to anticipated role changes and breakdown of relationships after predictive results were obtained seems to be a common occurrence (Codori & Brandt, 1994; Quaid & Wesson, 1995; Sobel & Cowan, 2000), and affected two of the participants from this study. Although it has been noted that breakdown of relationships occur after receiving a positive HD result, Decruyenaere (2004) found that some couples broke off the relationship for reasons that were unrelated to the predictive test results, such as communication problems, differences in values and personalities, differences in need for independence and financial problems. The experiences of the participants in this study support the findings of the Huntington's Disease Youth Organisation (2012 a) which suggest that individuals who tested positive for HD worry whether they will develop HD, about whether they will be able to or should have children or even about getting married. One of the participants in this study reported, for example, that he is anxious about entering a new relationship because he would need to tell his partner that they would need medical intervention in order to conceive one day. Erwin et al. (2010) conducted a study to investigate the discrimination experienced by individuals with HD in relationships. The findings suggest that 51.2% of the participants reported that they worried about future discrimination or stigma if they had to enter new relationships and the possibility that they may be treated differently by their partners because of their family history of HD. It is evident that relationships can be strained not only by the presence of HD, but even due to a family history of HD.

**5.2.1.5. children.** Another frequently reported challenge by the participants of this study is the issue about having children. Participants reported that they had to consider alternative methods of conception, because they did not want to pass on the HD gene to their children. One of the alternative methods that participants mentioned, in order to conceive, was to go through in vitro fertilization (IVF). However, this is very costly, as one cycle of IVF could cost between R25 000 to R38 000, with a success rate between 35% and 50% (Pregnant today, 2013). A study by Decruyenaere et al. (2007) revealed that 35% of their participants decided to not have children once they found out that they tested positive for HD. Reasons for not wanting children include the risk that the child might develop HD and parents might not be able to fulfil their roles as parents if they themselves develop HD (Downing, 2005). One participant in the current study chose not to have children in the future after she found out that she tested positive for HD. In addition to considering alternative methods of conception and changed views regarding having children, participants also experienced guilt because of the risk of having passed on the HD gene to their children. Quaid et al. (2010) identified feelings of guilt in participants with HD, however, these feelings were identified among individuals who knew that they were at risk for HD and still wanted children. However, in this study, participants that expressed feelings of guilt about having possibly passed on the HD gene to their children did not know HD was in their family until their children were a couple of years old. The last and probably the most extreme challenge reported in this study was that one participant and his wife chose to terminate their pregnancy when the foetus tested positive for HD. Kromberg et al. (1999) also reported that three of the seven participants in their study that evaluated Johannesburg's predictive and diagnostic programmes terminated their pregnancies when they found out that they carried foetuses with the HD gene. Terminating pregnancies are agonising and depressing for the

couples and support, empathy and ongoing counselling are required by these couples (Kromberg et al., 1999).

**5.2.1.6. *it's a monster.*** Participants also shared their intense emotions about how they felt about having HD. HD was described as a frightening condition, which evoked feelings that ranged from anxiety to hate within them. It is clear that this is a terrifying condition, because the symptomology of HD affects several aspects of an individual's life, i.e. physical, mental and social parts of life.

**5.2.1.7. *employment.*** One aspect of an individual's life that HD has a major impact on is employment. HD is a progressive brain disease where both neurological and psychiatric symptoms occur, and generally significantly affects an individual after 5 years of onset, which means few would be able to continue working after this time (Harper et al., 2004). A recent study conducted in Canada, Australia and the United States, which looked at discrimination experienced by individuals with HD, found that 6.5% of participants experienced employment discrimination (Erwin et al., 2010). Discrimination reported by participants included being denied a job, being fired, being covertly watched or being denied a promotion (Erwin et al., 2010). It should be noted that the participants in the study by Erwin et al. (2010) were asymptomatic, meaning they did not display any of HD symptoms yet but were at risk of developing HD in the future. It is important to evaluate discrimination in the workplace as employment is crucial for income, social security and social status (Harper et al., 2004). However, the participants in this study had to stop working due to their HD symptoms, such as memory, concentration and movement problems, which left them unable to complete tasks. According to South African law, HD is seen as a disability as the definition of a disability is "a condition caused by an accident, trauma, genetics or a disease which may limit a person's mobility, hearing, vision, speech, intellectual or emotional functioning"

(Department of labour, 2007, p. 1). If an employee presents with a disability, the employer is required by law to provide suitable and reasonable accommodations to help the disabled employee (Department of labour, 2007, p. 1). Reasonable accommodations may include workstation modifications, such as making it more wheelchair friendly, adjusting work schedules, adjusting the nature and duration of the employee's duties at work and reallocation of non-essential tasks or other modifications to how work was previously or currently performed (Department of labour, 2007, p. 1). It doesn't seem as if the employers of the participants that reported that they could not work anymore in this study made any special accommodations in order to prolong their employment with the company. Two participants reported for example that their cognitive symptoms worsened as the day progressed and both reported problems retaining work. A possible accommodation that would have been reasonable would have been to adjust their work schedule or work hours to try to accommodate these participants. One participant also reported that it was difficult to deal with her positive test results, while still committing to a full-time job. Guidelines set by the Huntington's Disease Society of America (HDSA) emphasise the importance of the timing of the testing process, as it is a stressful process and one should not consider starting the process during a stressful period, especially when the individual experiences a lot of stress at work (HASA, 2013). Work challenges are worth further investigation to determine the frequency and extent that these problems occur and to assess whether employers are following the labour laws and/or whether discrimination against people with HD occur.

**5.2.1.8. social support.** Another challenge that participants in this study faced was in terms of social support. Social support included their spouses/partners, their nuclear and larger families and support group meetings. Complaints about spouses included them being unsympathetic, intolerant at times, like for example when a participant perceived that his

spouse got irritated when he could not walk fast enough. Kaptein et al. (2007) reported that unsupportive behaviours by spouses can negatively influence a patient's well-being.

The second challenge, concerning social support, extended to the participants' larger families. The first challenge regarding the extended family was that some participants had false perceptions about family members with HD before they knew it was HD and, as a result, had judged them because of their abnormal behaviour. This was mainly because information about HD was scarce and family members with HD was kept a secret and not spoken of. Many adults, who were not told about HD and who did not have the opportunity to discuss it at home, felt angry, resentful, hurt and disempowered when they are informed about HD (Huntington's Disease Youth Organisation, 2012b; Klitzman, Thorne, Williamson, Chung & Marder, 2007). As a consequence, their anger can hinder coping with HD (Huntington's Disease Youth Organisation, 2012b). These adults may experience their family as untrustworthy and that the decisions they made in life was made without all of the information available (Huntington's Disease Youth Organisation, 2012b). It would have perhaps been useful to ask participants, who were not aware of HD in their family, whether they would have made different decisions, such as marrying and having children, if they were informed about their family history of HD before they made those choices. This should be considered for future studies in HD.

Staying with social support, disclosing a positive result to the family also seemed to pose as a challenge for some of the participants, especially when parents have to inform their children. Forrest-Keenan, Miedzybrodzka, van Teijlingen, McKee and Simpson (2007) implied that informing children at a younger age rather than older age, equips them to cope with their risk better. One of the participants chose to not tell her daughter that she tested positive. Her reason was that her daughter could only test herself at the age of 18, meaning



she would not be able to take any action once her mother disclosed her status to her. This could potentially be a problem when the daughter finds out that her mom has HD and she was never informed. Hennig (2003), the author of 'Talking to kids about Huntington's Disease', suggests that telling children about HD and the parent's HD status at a younger age will mean HD will become more normal for children as they grow up. When parents disclose their HD status, they should remember whether HD was kept a family secret for them and how they felt when information was kept from them (Hennig, 2003). Unfortunately, the topic of HD is not a once off discussion and should be revisited often in order to give everyone the latest information about HD (Forrest-Keenan et al., 2007; Hennig, 2003).

Another challenge that participants had concerning their families was that some of them worried about their family members with HD in terms of looking after themselves and being financially stable. One participant described how she was worried about her brother as he was experiencing difficulties at work because of his HD while having to support his family. The study by Bombard et al. (2011), which looked at individuals at-risk for HD and their concerns for themselves and their families found that 73% of individuals were concerned about family members experiencing genetic discrimination. Therefore, it is not uncommon for family members to stress about other family members with HD, although it is also a source of stress for individuals with HD themselves.

Another challenge that three participants reported with regards to their extended families, as part of their social support structure, was that some felt distressed by seeing their family members and how they deteriorated as HD progressed. This gave them a glimpse into their future and to some this was upsetting. Seeing family members with HD deteriorate is very difficult (Huntington's Disease Youth Organisation, 2012c). However, participants did not only report the deterioration of family members as sad, but also distressing because they

saw what would happen to them, a topic which has not been covered in other studies, according to my knowledge.

Emotional cut-off, such as denial, has also been noted as a problem in family members and spouses where an individual has tested positively for HD (Smolina, 2007). The avenue of denial could be a conscious effort for self-protection from devastating emotions, and in return offers the individuals comfort, relief and alleviation of pain (Evers-Kiebooms & Decruyenaere, 1998). Participants in this study mentioned that some family members were in denial about family members having HD. This hindered the coping of both the participant and the larger family. With the onset of HD, individuals have to face and deal with potentially burdensome and stressful information, which can be considered a traumatic event with emotional consequences, such as helplessness, depression and an uncertain future (Smolina, 2007). From this statement it is clear why individuals may choose to be in denial about HD. However, prolonged denial can be devastating as it does not allow the individual, who is in denial, to adapt to their reality and adequately prepare for future situations where family members progress with HD (Kessler & Bloch, 1989; Lowit & van Teijlingen, 2005).

The last challenge about the participants' families was that a third of the participants in fact viewed themselves as a current or future burden to their families. Previous studies also suggested these fears of being a burden (Johnson, Sulmasy & Nolan, 2007). According to Johnson, Sulmasy and Nolan (2007), the feelings of burden that chronically ill individuals have can have emotional, social, financial and physical aspects. However, physical aspects were more prominent in the current study as participants worried that they would need intensive care in the future, for example, personal hygiene and feeding regimens. It is clear that the participants from this study struggled with the possibility that they would need care in the future, which would either have to be done by their spouses or nursing staff. As a

result, some participants felt guilty about having HD, as they may require more care than was initially expected before they found out that they have HD.

The last form of social support that was reported as a challenge was the monthly support group meetings. Although the majority of the responses about the support groups were positive, one participant mentioned how the meetings can sometimes be emotionally draining and overwhelming because of having to listen to other's problems in addition to dealing with your own difficulties. Although no literature on HD and negative outcomes of support groups exist, a similar study has been performed using cancer patients and their significant others (Galinsky & Schopler, 1994). Galinsky and Schopler (1994) found that 25% of respondents in their study stated that they felt overwhelmed by the information and responses from other members when they joined the support group meetings. This frequency is lower in the current study, as only 1 of the 6 participants who are involved in support groups reported this to be a problem. As with the current study, Galinsky and Schopler (1994) note that this complaint was not very frequent and not very severe, however, it warrants attention. Facilitators of support groups should be trained in order to understand the factors that can impede group discussion and progress, as well as be informed about conflictual elements, which are normal during a group's development. (Galinsky & Schopler, 1994).

### **5.2.2. Meso-system**

The only challenge that was identified on the meso-level of Bronfenbrenner's Ecological model was the interaction that took place between the participants' partners/spouses and their family members with HD. This was described as a great concern for one of the participants as she felt that having her partner see her family members with HD might frighten him. To my knowledge, this has not been noted in previous studies regarding

HD. This may be because studies have either focussed on the individual with HD or family members separately, without acknowledging the interactions between partners/spouses and family.

### **5.2.3. Macro-system**

The next level where challenges were identified was on Bronfenbrenner's Ecological Model's fourth level, namely the macro-system. Challenges that participants encountered related to medical aid, insurance policies, finances, lack of HD facilities and a general lack of understanding of HD.

**5.2.3.1. medical aid.** The first challenge reported by participants on this level was that they found medical aid schemes very expensive because they had received a positive result on their predictive test. A third of the participants explained that if they wanted to claim for HD related medicine from their medical aid scheme, they had to be on the most expensive scheme, and that this was a very costly option to consider. For example, Discovery, a medical aid firm in South Africa that was specifically mentioned by participants, requires an individual who tested positive for HD to be on the Executive or Comprehensive plan before they could claim for treatment (Discovery Health Medical Scheme, n.d.) These plans range from R2335 to R3764 for a main member per month, whereas cheaper plans start at R556 per month (Discovery Health Medical Scheme, n.d.). Another South African medical aid, Libmed, also requires individuals with HD to be on their two most expensive plans (Liberty, 2014a), which are 'Traditional ultimate' and 'Complete Plus', costing each main member R4936 (Liberty, 2014b) and R3810 per month (Liberty, 2014c), respectively. This information highlights the problem that individuals with HD face with regards to medical aid schemes and having to choose the more expensive option in order to be covered medically.

**5.2.3.2. *life insurance.*** Life insurance policies emerged as another major challenge for the participants in this study. While life insurance became significantly more expensive for some participants once they received a positive predictive result, others struggled just to obtain a life insurance policy. In an international study by Bombard et al. (2010) that looked at genetic discrimination among individuals with either a positive predictive HD test result or at risk-individuals with a HD family history found that 14.5% were denied life insurance. Another problem that was identified in a similar study was that premiums were increased for individuals with or at risk of HD (Bombard et al., 2009). Momentum, a South African insurance company, will decline life insurance for an individual who already has the onset of HD symptoms (M. Alberts, personal communication, January 14, 2014). However, if the individual only has a family history of HD and has tested negative for the HD gene, then normal premiums will apply (M. Alberts, personal communication, January 14, 2014). The last scenario that would apply is if there is a family history of HD and the individual received a positive result, but is asymptomatic, then the following premium loadings apply: ages up to 39 have a 200%, ages 40 to 49 have 150%, and 50 to 59 have 100% (M. Alberts, personal communication, January 14, 2014). Another life insurance company in South Africa, namely Sanlam, is likely to decline life insurance for an individual with a positive test result if he/she is younger than 35 years old (R. Kraemer, personal communication, January 14, 2014). It was not clear whether older individuals with a positive test result will be able to acquire life insurance (R. Kraemer, personal communication, January 14, 2014). However, if there is a family history of HD and the person has not yet been tested, then a young person would have a premium loading of 150% whereas older individuals can have as little as 50% (R. Kraemer, personal communication, January 14, 2014). Life insurance therefore seems to be a challenge for individuals with HD both internationally and nationally.

**5.2.3.3. financial problems.** Whereas policies became more expensive for individuals with HD, almost half of the participants reported how HD had negatively affected their household incomes. One way in which HD had affected their income was that three of the participants had to stop working, which meant two of them were living on disability grants and one reported that he could not receive company benefits anymore, such as bonuses and shares. A person who wants to claim for a disability grant, from 2012, can get a maximum of R1200 per month, depending on the extent of the disability (Department of Social Development, 2014). This means that HD can limit an individual's income significantly. In other parts of the world, for example United Kingdom, an individual with a disability, such as HD, can receive up to 160 pounds a week (Disability living allowance for adults, 2014), which is considerably more than in South Africa. With little additional help from the government when disabled, it is clear why raised premiums for medical aid and life insurance pose as a challenge for individuals with HD.

**5.2.3.4. lack of HD facilities.** Unfortunately, another challenge that seemed to be frequently reported was the lack of HD facilities in the Western Cape, and the rest of South Africa. Care facilities are either necessary for the participants' family members who have already progressed far with HD or themselves once they have reached a certain stage with HD. This was concerning as a lack of HD facilities, which have specialised facilities and trained staff for HD, left participants worried and anxious about their future and where they might receive the proper care one day if needed. Unfortunately, there are no HD specific facilities in the Western Cape, or in the rest of South Africa. Compared to other countries in the world, the United States has 9 specialised caring facilities for individuals with HD, one which recently opened in 2012 that offers 32 beds for HD patients (Huntington's Disease Society of America, 2012). Although some countries offer specialised caring facilities,

according to literature, carers of family members with HD and individuals with HD themselves are not always entirely satisfied with the facilities (Etchegary, 2011; Kristjanson, Aon & Yates, 2006; Skirton, Williams, Jackson Barnette & Paulsen, 2010). In a Canadian study by Etchegary (2011), which looked at health care resources for individuals with HD, it was reported that where they had family members in care homes participants experienced that the staff were untrained for HD and that some of their family members had received inappropriate care because of this lack of training. Similar to the findings of the current study, Etchegary (2011) found that the participants in their study were worried about their future as there is a lack of caring homes for individuals with HD in Canada. Similarly, a study that looked at HD services in both United Kingdom and United States found that the most frequently reported complaint by carers about health care services was that there was a lack of community resources to help them with HD (Skirton et al., 2010). This complaint included that the staff in the care facilities were not properly trained to provide proper care for individuals with HD, that it was too expensive, too far away from home and that carers struggled to get their loved ones accepted into such a facility (Skirton et al., 2010). Facilities are crucial because as patient dependency increases, more support is needed because both patients and carers display increased distress symptoms and decreased quality of life (Kristjanson et al., 2006). Patients who receive more palliative care and tailored services are more satisfied than those patients who did not received such special care (Kristjanson et al., 2006). It is clear from this discussion that there is a great need for HD facilities with medical staff who specialise in the treatment of individuals with HD in the Western Cape and greater South Africa in order to relieve stress and anxiety in both individuals with HD and their family members.

**5.2.3.5. *lack of understanding.*** Not only are facilities absent in South Africa but there is also a lack of understanding of HD among the general public. This was reported by almost all of the participants as a major challenge. This lack of understanding negatively affected the participants as some stated how they felt lonely and misunderstood, while others reported how their family members had been maltreated because doctors and nurses were not properly informed about HD. One of HASA's aims is to raise awareness in South Africa about HD, however, this organisation is still relatively new as it started in 2004, compared to the United States HD organisation, which started in 1968 (HASA, 2013 HDSA, 2012). Unfortunately, a lack of understanding of HD also extended to the participants themselves, as many of them did not know what HD was when their family members were diagnosed with HD, which made it difficult to determine why they were displaying certain peculiar behaviours.

#### **5.2.4. Chrono-system**

The last level of Bronfenbrenner's ecological model is the chrono-system, which consists of two sub-themes, namely symptom watching and the progression of HD.

**5.2.4.1. *symptom watching.*** Symptom watching is a common occurrence among individuals who are at risk of developing HD. This refers to them becoming anxious if they notice certain behaviours or actions, such as forgetfulness or dropping objects, similar to what their affected relatives displayed, and it involves wondering whether it is the onset of HD (Division of Human Genetics, 2013). Participants reported how they got anxious when they knocked things over, when their feet suddenly jerked, when they dropped objects or tripped. They then went on to report that it was difficult to differentiate which movements or behaviours were as a result of HD and which were normal actions. It is suggested that if symptom-watching produces a lot of anxiety then the individual should visit a genetic counsellor or a neurologist (Division of Human Genetics, 2013).



**5.2.4.2. progression of HD.** The last sub-theme, pertaining to the chrono-system, was that participants reported how the progression of HD was a challenge for them. This was because participants acknowledged the fact that their HD would progress and their condition would worsen. . This caused anxiety for some and they stressed about how they would be able to provide for their families financially, as well major life changes that would need to occur, such as moving houses, if their HD progressed to that point. Unfortunately, the nature of the progression of HD is unpredictable and unique for each individual, making planning for the future very difficult (Klager, Duckett, Sandler & Moskowitz, 2008). As well as being unpredictable, the progression of HD has no periods of remission, which was why participants who already displayed symptoms of HD felt anxious as they knew that their HD would only progress (Huniche, 2011; Kromberg et al., 1999).

### **5.3. Supports/resources**

The next major theme, which also speaks to one of the aims of this study, was the resources and/or supports that participants found useful to cope with their HD. As previously mentioned, supports were identified on all of the levels of Bronfenbrenner's Ecological System's Theory, except on the exo-system. The reason for this is that this particular study focussed on the experiences of the individual with HD, whereas the exo-system is concerned with social settings in which the individual is not actively involved, such as the individual with HD's spouse and that person's work environment.

#### **5.3.1. Micro-system**

On the microsystem of Bronfenbrenner's Ecological System's Theory, seven supports/resources were identified as being valuable to the participants in the quest to cope

with HD. These supports and/or resources were: knowledge about HD, counselling, medication, attitude, employment, social support and the testing process.

**5.3.1.1. knowledge about HD.** The first resource that participants reported as helpful was all the information about HD, which was available from books, doctors and the internet. There are several books about HD, both novels and factual. Books about HD were reported to improve the participants' understanding of their own HD, as well as their family member's with HD, and why their family members were displaying certain behaviours. It also left the participants feeling less isolated because as they read about other families with HD they realised they were not alone. Other sources of information, such as the internet with web pages dedicated to HD, such as 'HD Buzz' were also reported to be important resources for finding the latest updates and factual information, which was written in relatively simple terms so that anyone could understand it. Other studies have also found that information is essential for coping with the diagnosis and progression of HD (Dawson, Kristjansen, Toye & Flett, 2004). However, there seems to be an important balance between the timing, type and amount of information (Dawson et al., 2004). It was found that too much information following the diagnosis of HD was experienced as stressful (Dawson et al., 2004). According to Dawson and colleagues (2004), acquiring information, which was readily available and close by, was vital for individuals with HD and carers, and the main source of information in Australia was the different branches of the Australian Huntington's Disease Association (Dawson et al., 2004). Participants in the Australian study reported that they contacted their closest association for all the current information, which included the latest research updates, resources and services available for HD (Dawson et al., 2004). Participants from the current study used similar types of information, gained from HASA support group and internet sources to make informed decisions about their future, such as having to put policies in place

and to make decisions about how they would like to be cared for once they reach an advanced stage of their HD. It is evident that knowledge about HD served many purposes for participants in our study; however, most of their information was from international sources, such as the internet and books.

**5.3.1.2. counselling.** Another important resource that helped many of the participants to cope with HD was counselling sessions. Participants reported how talking to someone about HD and their challenges was helpful, as they knew everything that was said in those sessions was confidential, and in addition, they felt supported by their mental health practitioner. Psychotherapy, as provided by a psychiatrist, a psychologist or a clinical social worker, can help patients manage their behavioural problems, develop new coping strategies, manage expectations during the progression of HD and facilitate effective communication strategies among the rest of the family (Band Back Together, n.d.; Indiana University Neuroscience Centre, n.d.). Unfortunately no studies have been performed, to my knowledge, examining the benefits of psychotherapy among HD individuals. However, a pilot study, conducted with terminally ill cancer patients, found that with a psychotherapy group intervention programme feelings of hopelessness and a desire to hasten death diminished (MacReady, 2003). The study also concluded that participants were less anxious and depressed and experienced greater spiritual well-being (MacReady, 2003). It is possible that group therapy could have similar positive effects on HD patients.

**5.3.1.3. medication.** In addition to knowledge about HD and counselling, participants reported that medication to treat HD symptoms was an important resource to manage their HD. It should be noted that there are no cure for HD and the nature of pharmacological treatment is generally symptomatic. Treatment is normally limited to anti-dopaminergic agents, which are prescribed for movement disorders, and anti-depressants for mood

disorders, while unfortunately treatment for cognitive problems remains unavailable (Venuto, McGarry, Ma & Kieburztz, 2012). The most frequently reported medications by participants in this study were sleeping tablets, anti-depressants and medication for their choreic movements. Tetrabenazine is the only FDA-approved drug for the treatment of choreic movements and clinically improves chorea scores (Frank & Jankovic, 2010; Gudesblatt & Tarsy, 2011). Participants used this to control their chorea, especially at night, to help them to sleep with less movement. Sleeping tablets are also often prescribed to individuals with HD, with Trazodone being prescribed often as it has positive effects with low side effects (Zhou, Jung & Richards, 2012). Lastly, medication for depression and aggressive behaviour is also prescribed, which may include Citalopram and Sentraline respectively (Roos, 2010). The treatment of individuals with HD must be tailored to each individual, because HD signs and symptoms usually differ in every person and may change over time (Roos, 2010).

**5.3.1.4. coping.** Another important resource for individuals with HD was the coping mechanisms that they utilised. Several attitudes were described as being helpful when trying to cope with HD, one being a positive attitude. Etchegary (2009) found that a positive attitude was employed by individuals with HD as they knew that there is no cure and that options for treatment are limited. Thus they had to generate other solutions to cope with this condition, one being a positive attitude (Etchegary, 2009). A positive attitude seemed to give individuals with HD a sense of control over a disease which offers almost none (Etchegary, 2009).

Another coping mechanism that was deemed important was acceptance of the disease, not only by the participants themselves but also their family members. Acceptance, which has been studied as a coping mechanism in individuals with HD, whether in the prodromal stage or after onset, has been rated as the most frequently used strategy in order to cope (Downing,

Williams, Leserman & Paulsen, 2012; Helder et al., 2002; Kaptein et al., 2006). Acceptance of a chronic disease can contribute to the patient's emotional stability, presenting them with an opportunity to reflect about alternative ideas about how to cope when they are left to confront new challenging situations (Helder et al., 2002). In the case with HD, which is an unpredictable, uncontrollable and to date, incurable disease, acceptance of HD and the consequences thereof is vitally important (Helder et al., 2002).

The most common point of view that was reported by participants in this study was that they could pass away from something unrelated to HD, such as a car accident or another condition. Although this was a prevalent response in this particular sub-theme, previous work on HD has not mentioned this, to my knowledge. It is just acknowledged that individuals who do in fact have HD may pass away before the onset of symptoms (Nordqvist, 2009). However, this should perhaps be emphasised more to individuals with HD, as it seemed to make the participants less preoccupied with HD, as it may not even develop further or at all in their life time.

Another type of coping mechanism that was reported by participants as useful was to make the most of every day. This is in line with other studies (Decruyenaere et al., 2004; Williams et al., 2010).

An interesting attitude that was noted during analysis of the participants' responses was how they compared their situation, of having HD, with other people, for example those who had financial difficulties or had another condition such as cancer, and felt that their situation was better. This can be explained in terms of Festinger's (1954) theory of social comparisons, where individuals learn about their opinions and abilities by means of comparing themselves to others. Not only do social comparisons serve an informational function, but they also serve an important motivational purpose, being crucial for both affect

and self-esteem (Festinger, 1954). Wills (1981) expanded on this theory and included the concept of 'downward comparison', which is when an individual looks at another individual or comparison group that the individual considers to be worse off. This serves to dissociate the individual from others and to make the individual feel better about him/herself or his/her personal situation (Wills, 1981). Downward comparisons can improve one's subjective well-being (Wills, 1981). For example, downward comparisons has been noted in patients with breast cancer who described themselves as being better off than other patients who were less fortunate than themselves (Wood, Taylor & Lichtman, 1985). Social comparisons have also been noted among individuals with HD in order to help them cope with the risk of having HD or the illness itself (Etchegary, 2009). However, the social comparisons that were noted in the study by Etchegary (2009) differed, as they mostly included participants comparing themselves to their deceased family members who had HD and found it comforting to know that those family members had a late onset age of HD and believed that they would too only show HD symptoms much later in life. As previously stated, social comparisons in the current study mainly included participants comparing their situation of having received a positive predictive result to other stressful situations, such as poverty or living in a war zone, and found that their situation was less severe. Participants who used social comparisons in this study tended to still be in the prodromal or early stages of HD.

The last coping mechanism that seemed to work for participants was taking it day-by-day. Many guidance books about HD and encouraging posts on online support message boards shared by individuals with HD themselves, suggest taking it one day at a time (Dawson et al., 2004; Kelly, 2006; Quarrell, 2008). It is evident by the range of responses that there is no specific attitude needed to cope with HD; it depends on the individual what he/she prefers.

**5.3.1.5. employment.** Contrary to what was previously mentioned about the work environment as a challenge for some, others found their work to be a resource and coping method. Two benefits of employment was described by participants, one being that a job kept the participants busy during the day, leaving little time for them to think about their HD. The second is that the participants had an additional source of support, which consisted of their colleagues and employers at work. Although benefits of employment in individuals with HD has not been studied, it has been extensively reported concerning another neurological condition, namely Multiple Sclerosis (MS). Benefits reported from employment among MS participants include the above mentioned as well as: providing them with a salary to pay for medication and playing an important role in their self-esteem as well as socialisation (Johnson et al., 2004). From the participants' responses during the interviews, it is evident that employment served many positive roles. Therefore, employers should try to be supportive where individuals with HD are concerned, as these individuals view their employment as incredibly beneficial.

**5.3.1.6. social support.** Another support that was part of the participants' micro-system was the various social support structures which gave them the support they needed to cope with HD. Social support included spouses, friends, family and support groups. Spouses seemed to be the most frequently reported form of social support that participants found most encouraging. This was because spouses seemed to be informed about HD and what will happen in the future, they did not leave the participant after the positive predictive test or when they started to present with symptoms of HD, and that they treated the participants as normal individuals. Partners or spouses who are informed about the risk of HD and what it entailed seem to react better to the effects of genetic testing (Smolina, 2007). It is imperative

to acknowledge the important role that spouses play in the individual with HD's experiences as it seemed to provide them with a stable source of support.

Friends, however, were not experienced as a major source of additional support. Many of the participants reported how they did not share their family history of HD with their friends. Only two participants reported that their friends acted as a support for them. This may be because many individuals with HD reported feeling discriminated against by their friends once they divulged their positive status to them, by receiving negative comments and changed behaviour and treatment towards them (Erwin et al., 2010).

Although friends were not deemed a major source of support, the participants' larger family seemed to provide much needed support. HD had the ability to bring families together to strengthen bonds and to create a safe space for discussions around the topic of HD. This is in-line with findings of other studies (Duncan et al., 2008; Vamos, Hambridge, Edwards & Conaghan, 2007; Williams et al., 2010). Although it was previously stated that some participants found it difficult to watch family members regress as HD progressed, others found family members with HD to be a support as they knew what to expect in their own future. It was reported that watching how family members handled their HD was inspirational and made them calmer about the future. This has gone unnoticed in the current literature. Participants also stressed the importance of family members offering care, as some of them needed help with preparing of meals and other daily activities. Most of the caring of individuals with HD became the responsibility of their family members (Skirton et al., 2010). It is important to note that individuals with HD appreciated their family's care and that it was viewed as an important source of support for them.

The last form of social support that was reported by participants as being helpful in their journey to cope with HD was the support groups offered by HASA. These meetings



were deemed important as it offered them an opportunity to come into contact with others who had the same condition, which meant that participants felt less isolated while having to deal with HD. Professionals who joined these sessions also shared valuable advice and could update the members on the latest research and findings. Both professionals and other members inspired participants to help them cope. Lastly, support sessions seem to encourage participants to talk about their experiences, thus creating a network of information where advice is shared. All of these benefits from a support group are well documented in previous literature (Family Caregiver Alliance, 2003; Galinsky & Schopler, 1994; Walker, 2007). It was not expected that new benefits would arise from these support groups during the interviews with the participants from the current study; however, it was more important to evaluate the perceptions of members of this group, as many of the participants attended these support group meetings regularly and the group was relatively new. Overall, the majority of the responses pertaining to these support groups, offered by HASA, were that these sessions were very valuable to the participants. This is useful to know as these meetings are relatively new, and have never been evaluated. It seemed that social support, consisting of spouses or partners, friends, family and social support meetings, played a very integral part in the participants' lives and their ability to cope with HD.

**5.3.1.7. testing process.** Contrary to what was previously stated, the last resource in the participants' micro-system is the testing process, which lead to a positive result on their predictive test for all of the participants in this study. The testing process was helpful for two reasons, the first one being because of the staff involved and second, what the results meant to the participants.

The medical staff that were involved consisted of genetic counsellors, psychologists, neurologists and genetic nurses. One of the reasons why they were deemed helpful was

because of all the information received, especially by the genetic counsellors who explained how HD is passed on and what HD is. This is in contrast to an article that found that only 3% of 1065 participants thought that a genetic counsellor was the best source of information regarding HD (Stern & Eldridge, 1975). However, the study by Stern and Eldridge (1975) was conducted almost 40 years ago and it is likely that genetic counsellors are now more informed about the critical role they play in providing their patients with knowledge about the disease. It was important that staff used simple terms and explained medical concepts. Another reason why the staff were seen as helpful during the testing process was because participants felt that the process was not rushed and that it was very thorough. This is because, according to the guidelines followed by the Groote Schuur genetic testing unit in Cape Town, the whole process consists of six sessions (Division of Human Genetics, 2008). Results will only be disclosed to the participant at the fifth meeting, which is a minimum of 16 weeks after the initial meeting (Division of Human Genetics, 2008). Also, a follow-up session is also included; approximately a month after the individual received his/her results in order to assess how that person is coping (Division of Human Genetics, 2008). Lastly, the medical staff was seen as a support because they require the individual who is going through the testing process to be accompanied by a friend or family member for additional support. Participants described how it helped them through the process as they were not alone during this stressful period.

The second component of why the predictive test was helpful to participants was the results themselves. All 12 participants had undergone the predictive test and tested positively, as this was an inclusion criterion for this study. None of them regretted the decision to know their status. This confirms that the testing process can be beneficial to individuals with HD. Their results meant that they could plan more adequately for the future, especially where their

will and insurance policies were concerned. Participants also displayed a greater appreciation for life and all that it had to offer, including opportunities and relationships. Lastly, participants reported how their anxiety had decreased and almost diminished after they learnt about their positive status. This is all in accordance with other studies that have examined predictive testing in individuals with HD (Gudesblatt & Tarsy, 2011; van 't Spijker & ten Kroode, 1997; Wiggins et al., 1992; Williams et al., 2010). Although, as previously mentioned, the testing process was also described as a challenge for some, as participants had to travel great distances for the testing and felt that the process was too long. However, overall, it was felt that more benefits than difficulties were experienced. The testing process could provide help for an individual who is at risk for HD, especially if the individual is anxious about his/her unknown status.

### **5.3.2. Meso-system**

There was only one support identified on meso-level of Bronfenbrenner's Ecological model. As previously stated, the meso-system is concerned with those interactions taking place between the micro-systems in one's life, such as the individual's partner and workplace. The support that was identified on this level was the idea that the participants' spouses or partners could observe and interact with their family members who had already shown HD symptoms. This means that partners and spouses were not disillusioned about the progression of HD and that they may very well face the same future one day. Another reason why the interaction between spouses/partners and family members with HD was seen as a support was because of the fact that the participants could not hide HD from them; participants had to deal with their condition within the relationship also. To my knowledge, this has not been cited in other literature. This may be because most of research regarding HD

is concerned with new advances in science instead of the qualitative experiences of individuals with HD and their families.

### **5.3.3. Macro-system**

Several resources and supports were also identified on the macro-system. These included medical aid, life insurance, a cure for HD, HD facilities, religion and grants.

**5.3.3.1. medical aid.** The first resource identified on the macro-level, as reported by participants, is the benefits that a medical aid provides individuals with HD. Although some participants complained that it was very expensive to have a medical aid if you have HD, as most medical insurance companies required the individual to be on the most expensive plan, others described how it was comforting to know that they were covered by a medical aid. However, it is not necessary for individuals with HD, who have not yet started to show symptoms, to be on a plan that specifically covers costs of HD. It is possible to up-grade plans at the end of each year, which is known as their open period (Discovery Health, n.d.). This is the period when members can change their medical plans to a more comprehensive one where more medical conditions are covered with additional benefits. This means that individuals can be on a cheaper plan, with fewer benefits, and once they require medication for their HD symptoms, they can up-grade to a plan which would cover their HD related medication. A medical aid was reported to be beneficial when participants would have to start claiming for HD related medicine, such as for their movement disorders. Other future claims, which medical aids would be beneficial for, would be when participants develop pneumonia, which is the leading cause of death in individuals with HD (Heemskerk & Roos, 2011). Pneumonia, as prescribed by the Council for Medical Schemes (n.d.), must be covered by all medical schemes in South Africa. This means that if these participants develop pneumonia and have a medical aid then they will not have to worry about hospital expenses. This is

considered to be a significant resource for HD as individuals do not have to stress about their medical expenses, which would be as a result of HD one day, when they are sick in hospital.

**5.3.3.2. *life insurance.*** Life insurance emerged as another important resource for the participants in this study. Similar to predictive testing, life insurance was also previously portrayed as a challenge for a few participants. According to both Momentum and Sanlam, for individuals who are at risk for HD, it is best to take out the life insurance policy before going through with the predictive testing. This is because if the individual tests positive for the HD gene, then it is most likely that life insurance will be denied for him or her (M. Alberts, personal communications, January 14, 2014; R. Kramer, personal communication, January 14, 2014). However, if the person only has a family history with HD and has not yet been tested, then that person's premiums will have a loading of between 50% and 200%, depending on the age, usually the younger the person is, the higher the loading (M. Alberts, personal communications, January 14, 2014; R. Kramer, personal communication, January 14, 2014). It is also unlikely that an individual with an HD family history will be allowed to take out life insurance with a disability cover, meaning the policy will not ? pay out if the person cannot work anymore because of their severe HD symptoms; the policy will only be paid out when the individual with HD passes away (M. Alberts, personal communications, January 14, 2014; R. Kramer, personal communication, January 14, 2014). However, it is important to state that each individual's premium will differ with each insurance company. It is also advised that a person with HD talks to the medical underwriter team of each life insurance company, as they will be able to advise each individual about their premium loadings and whether they will likely be accepted or denied life insurance.

**5.3.3.3. *cure.*** The hope for a possible cure in the near future was also described as a support for participants, as new scientific breakthroughs were encouraging for themselves

and also for their children. Participants who had not yet started showing movement symptoms of HD described how they were hopeful that a cure would be found for HD by the time they showed symptoms. Others reported how their decision to have children without medical assistance, such as in vitro fertilisation so that the foetus could be tested for HD, was based on their hope for a cure by the time their children will start showing symptoms. However, hope for a possible cure for HD has been related to a high score for mental disengagement, such as turning to work or other substitute activities in order to distract oneself, or daydreaming about other things than HD (Helder et al., 2002; Kaptein et al., 2006). However, participants in the current study rather described their hope as a result of all the new scientific findings. Research in HD is multi-disciplinary and some of the fields include: basic neurobiology, which studies the HD gene in order to understand how it causes the disease, clinical research, which focuses on treatments of clinical trials and animal model studies, in order to mimic the disease in mice to test potential treatments (National Human Genome Research Trial, 2011; Rose & Russell, 2013). Although scientific findings are encouraging, it is important to not to have unrealistic expectations about scientific trials that may take years before coming into effect.

**5.3.3.4. possible HD facilities.** A more concrete potential source of support for the participants of this study is the long-term plan to up-grade Valkenberg Hospital, in order to include a specialised caring facility for individuals with HD. Valkenberg Hospital, a psychiatric facility in the Western Cape, is said to be upgraded by the end of 2016, with an additional 92 beds and 17 new buildings (Fokazi, 2012). According to Dr Baumann, a psychiatrist at Valkenberg, a specialised unit for HD will be included in the upgraded Valkenberg, which will offer respite and long term-care beds for individuals with HD (HASA, 2013). It seems as if 12, possibly 24 beds will be allocated solely for the HD facility

(HASA, 2013). Although it will take probably 10 years to complete this facility, knowledge about this gave participants a sense of relief as they knew they would have a place to go to if they progressed to a stage where they needed more intensive care, as well as a place they could take family members too. This made the participants less anxious about the future. This shows the dire need for proper care facilities in the Western Cape, as well as other provinces.

**5.3.3.5. religion.** Religion was the second last support that emerged on the macro-system. Similar to the findings of this study, religion is not often reported as a form of support among individuals with HD (Helder et al., 2002; Kaptein et al., 2006). In the study by Helder et al. (2002) that reported coping mechanisms of individuals with HD, religion was the second least frequently reported coping mechanism used. Although the minority of the participants in this study reported religion to be a source of support, these individuals made it clear that it contributed significantly to their coping with HD. Aspects of religion that participants focussed on in this study and in the study by Helder et al. (2002) differ greatly. For example, using religion as a coping mechanism in the study by Helder et al. (2002), according to the COPE inventory, included: trusting in God, seeking God's help, finding comfort in religion and praying more. However, statements of participants in this study, regarding their religion, comprised of: religion giving perspective about HD, a person's body is a temporary entity that will be disease-free when in heaven, and religion served a motivational purpose in order to make peace with HD and to give individuals a purpose in life. Although not a frequent form of support for individuals with HD, to some it is an invaluable coping mechanism, which serves many purposes.

**5.3.3.6. grant.** The last resource identified on the macro-system is the disability grant that two of the participants received. The disability grant is given to individuals who are unable to continue working because of a disability. As of 2012, the disability grant is R1200

(Department of Social Development, 2014). In addition to a disability grant, if the individual is unable to look after himself/herself anymore and requires someone else to look after them, then they are eligible to apply for a grant-in-aid (Department of Social Development, 2014). It is only an additional R285 per month, but is free to apply for (Department of Social Development, 2014). Both the disability grant and grant-in-aid can be paid electronically into the individual's bank account (Department of Social Development, 2014). This is convenient for individuals with HD because they generally have trouble walking and travelling because of their movement and cognitive problems. In addition to a disability grant, individuals who are disabled and cannot work anymore can apply for a housing subsidy (Department of Human Settlements, 2013). This would entitle the individual to R96362.00, if their monthly income is below R3500 a month, to put towards their house purchase (Department of Human Settlements, 2013). An additional set disability variance amount will also be given to the individual in order to pay for extra features for the house to make it more disability-friendly, such as a wheelchair ramp (Department of Human Settlements, 2013). Although the housing subsidy is available, none of the participants mentioned that they have made use of this. Individuals with an income below R3500 should be informed by genetic counsellors if they are eligible to apply, as this could help with mobility around the house. However, one of the requirements to qualify for this subsidy is that the person must have a healthy mental state (Department of Human Settlements, 2013). It is difficult to determine whether individuals with HD will qualify for this, as they often show cognitive and psychological symptoms of HD. The UK also provide disabled facility grants, in order to assist financially in the adaptation of homes of disabled people so that they can continue to live there (Inch, 2013). A disabled facility grant varies from 25 000 to 36 000 pounds, as a once off settlement, depending on where you live in the UK, and does not have the inclusion criteria of a particular mental status (Inch, 2013).



#### **5.3.4. Chrono-system**

The last level of Bronfenbrenner's Ecological Theory is the chrono-system, which represents the changes that happen over time in the individual's life. One such change that was identified on this level was that participants described how they would have time to adapt to the changes as their HD progressed. This is because HD can progress for up to 15 years with no periods of remission for 10 to 15 years (Huniche, 2011; Kromberg et al., 1999; Quarrell, 2008; Williams et al., 2011). This means that the progression is usually slow and no sudden or drastic changes are experienced. This gives the participants time to gradually change their lifestyle in order to adapt to their situation. One adaptation reported by participants was the fact that they would need to move houses, whether they developed HD symptoms or not, but because they were getting older. Knowing that changes, concerning HD, would be slow, participants felt that they could cope with their future.

#### **5.4. Limitations and strengths**

The first limitation is that since it was an exploratory, qualitative study, the sample was small and homogenous, including only relatively privileged individuals residing in the Western Cape. Consequently, it is not possible to make generalisations from this study to, for example, individuals with relatively low levels of formal education or to those who come from a socio-economically underprivileged backgrounds, as one would be able to do in the case of a large-scale quantitative study using random sampling. However, according to Lincoln and Guba (1985), one is not supposed to be able to generalise from a qualitative study. As a suggestion, further research should triangulate methods, which should include both quantitative and qualitative techniques, as both quantitative and qualitative studies in SA concerning HD are required.

A second limitation of this study is that some of the accounts during the participants' interviews included experiences which dated almost 15 years ago, especially about the testing process. These accounts may be inaccurate because of all the time that had lapsed in between the test and the interview. Experiences concerning the medical staff and their reactions towards their positive results may differ from what participants remembered. However, the literature on predictive testing suggests that it is a critical and sensitive period in one's life and it was therefore included in this study.

Third and lastly, the themes that emerged during thematic analysis could have been discussed with the participants from this study. Although transcripts were sent back to the participants in order for them to check the accuracy of the interview, a focus group with all of the participants could have been arranged, in order to see whether they agree or do not agree with the themes that emerged. This would have served as an additional method of checking. Unfortunately, this was difficult to achieve because a number of the participants travel with great difficulty due to their motor symptoms and because of time constraints.

As for the strengths of this study, this is the first of its kind in a South African context. This means that the experiences of the participants specifically with regard to the health care systems, policies and government aid systems, in South Africa, were explored as the study looked at what supports and challenges were associated with HD. Other studies regarding HD in South Africa have usually focussed on either the epidemiological or genetic nature of the condition. This study served to narrow the literature gap about the psychological experiences that individuals with HD experience in South Africa.

Secondly, as it examined the challenges and supports/resources that individuals with HD experience in South Africa, it has formed the basis for interventions for these individuals. It is our hope new studies will evolve from this new literature regarding HD, such as

interventions which are specifically designed for individuals with HD within the South African context.

### **5.5. Future recommendations**

As this was an exploratory study, the value of this study is considerable as it has attempted to take the first steps in order to understand the nature of the experiences of individuals with HD in a South African context. However, given that sampling for the study was convenience sampling of individuals with HD, it is essential to replicate the study with a more representative sample, in terms of socio-economic and cultural groups. In addition, this was a qualitative study, which used in-depth interviews to gain access to the experiences of this small group of individuals. However, quantitative studies should be conducted, with the focus on psychological distress, such as depression and anxiety, life satisfaction and coping mechanisms. Alternatively, studies should also investigate the experiences of caregivers of HD patients and examine the challenges and supports that they experience.

### **5.6. Concluding remarks**

The study aimed to explore the unique experiences of individuals with HD in the Western Cape, South Africa. This was necessary as knowledge of the experiences of these individuals in a South African context was absent in the current literature. In total, 12 participants with a wide range of physical and mental functioning were included in the current study.

Two main themes, which were challenges and resources, emerged during data-analysis of the transcripts. The first theme, namely challenges, refer to difficulties that participants experienced as a result of their HD. These challenges consisted of several sub-themes and were the following: triad of symptoms, sleep problems, testing process,

relationships, it's a monster, employment, social support, partners and family, medical aid, insurance policies, finances, lack of HD facilities, lack of understanding, symptom watching, and progression of HD. These sub-themes emerged across all the different levels of Bronfenbrenner's Ecological System's Theory, with the exception of the exo-system. Secondly, a number of resources were identified as being helpful in order to help participants cope with HD and included the following sub-themes: information, counselling, medication, coping, work, social support, testing process, partners and family, cure, possible facilities, religion, disability grant, life insurance, medical aid, and adaptation over time. As with challenges, only the exo-system was not represented during analysis.

The majority of the sub-themes were found in previous literature regarding HD. However, several unique challenges and resources were identified in this study. The first one, being a challenge, was the interactions between the participants' spouses and family members with HD. It was seen as a challenge because participants felt that, by having their spouses or partners observe their family members with more progressed HD, might frighten them and make them weary of the future. As previously mentioned, this may not appear in the literature because HD studies usually focussed on either the individual with HD or family members with HD separately. Another unique challenge to the South African context was the little additional help from the government, in terms of disability grants and facilities, which made medical aid and life insurance costly expenses. The last unique challenge was the lack of HD specific facilities in South Africa at the moment, as there are currently none. This should be addressed by the government, by providing facilities for these individuals, in order to alleviate anxiety about future care among individuals with HD.

Concerning supports, participants from the study reported that watching how their family members handle their own HD was inspirational and calmed them about their future.

This has gone unnoticed in the HD literature. Lastly, the important role that social support, in terms of spouses/partners, friends, family and support meetings, played in the participants' lives and how it helped them to cope with HD was evident. This has not been emphasised in other literature.

Although several limitations were mentioned, such as the small sample that was recruited from only one province in South Africa, although applicable in this situation, and not discussing the themes with the participants before the final report was generated, these could be addressed with subsequent studies. The strengths of this study, being the first of its kind in South Africa and having produced invaluable information for interventions, should be emphasised.

The study was the first to explore the experiences of individuals with HD in a South African context. It demonstrates that, although these individuals with HD face several challenges due to their condition, they also make use of several resources to help them cope with this neurological condition. Lastly, the findings that emerged from this study contribute to raising awareness about the experiences of individuals living with HD and will serve as a valuable foundation for tailor-made interventions for these unique individuals.

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## Appendix 1



1 February 2013

Dear Ninon Joubert

We would like to hereby invite you and your supervisor, Chrisma Pretorius, to join us at our Huntington's Disease Support Group which will be held on 7 February 2013.

We are looking forward to learning more about your research study and how we as people affected by Huntington's Disease can participate and get involved.

Warm Regards

Jessica Selfe  
DIRECTOR: HASA

## Appendix 2

### PARTICIPANT INFORMATION LEAFLET AND CONSENT FORM

**TITLE OF THE RESEARCH PROJECT:** The lived experience of individuals living with Huntington's disease in the Western Cape, South Africa

**REFERENCE NUMBER:** S13/04/087

**PRINCIPAL INVESTIGATOR:** Ninon Joubert

**ADDRESS:** 9 Brenton Crescent, Belvedere Estate, 7550

**CONTACT NUMBER:** 072 545 6678

You are being invited to take part in a research project. Please take some time to read the information presented here, which will explain the details of this project. Please ask the study staff or doctor any questions about any part of this project that you do not fully understand. It is very important that you are fully satisfied that you clearly understand what this research entails and how you could be involved. Also, your participation is **entirely voluntary** and you are free to decline to participate. If you say no, this will not affect you negatively in any way whatsoever. You are also free to withdraw from the study at any point, even if you do agree to take part.

This study has been approved by the **Health Research Ethics Committee at Stellenbosch University** and will be conducted according to the ethical guidelines and principles of the international Declaration of Helsinki, South African Guidelines for Good Clinical Practice and the Medical Research Council (MRC) Ethical Guidelines for Research.

#### What is this research study all about?

- This study aims to investigate the challenges that are faced by individuals living with HD. This study also aims to investigate the supports and/or resources that help individuals living with HD to cope.
- It is important to conduct this research because HD is a debilitating lifelong disease, which includes physical and psychological symptoms. A few studies have examined the living experience and resources of individuals with HD but no studies of this nature have been conducted in South Africa. Knowledge of this nature can inform the design and implementation of interventions for individuals living with HD.

- The study will be conducted at your home or at a location that suit you. At least 15 individuals will be included in this study. The data collection will start with the completion of a biographical questionnaire. This will be followed by a semi-structured interview, consisting of questions about your living experience with HD. This interview will last between 60-90 minutes. If you agree, the interview will be tape-recorded so that the interview can be transcribed verbatim to help with the process of data analysis.

### **Why have you been invited to participate?**

- You have been invited to participate in the study as you have been identified as an individual living with HD. You also expressed an interest to participate in this study at the HD support group meeting that was held in Claremont on the 7th of February 2013.

### **What will your responsibilities be?**

- Your responsibilities for participating in this study involves that you take part in a once off semi-structured interview lasting from 60- 90 minutes to talk about your experiences about living with HD. To discuss and verify the results of this study, a focus group, which will include the other participants of this study, will be organised after the completion of all the interviews. If you would like to participate in this, we will notify you of the date and location.

### **Will you benefit from taking part in this research?**

- You will not directly benefit by taking part in this study. However, the findings of this study might be published as a scholarly article in a peer reviewed journal. Publication of these findings will hopefully be the first step towards bringing awareness about the lived experience of Huntington's disease in a South African context and might stimulate further research in this field.

### **Are there any risks involved in your taking part in this research?**

- The only foreseeable risk that participants may experience is emotional distress and/or discomfort during the interview due to the personal nature of the interview. If this is the case, I will refer the participants to my supervisor, Dr. Chrisma Pretorius, a Counselling Psychologist, who will refer you to the appropriate health care professionals. Her contact details are: 021-808 3453 or [chrismapretorius@sun.ac.za](mailto:chrismapretorius@sun.ac.za)



**If you do not agree to take part, what alternatives do you have?**

- This study does not include any form of treatment, so no alternatives will be offered.

**Who will have access to your medical records?**

- Any information that is obtained in connection with this study and that can be identified as you will remain confidential and will be disclosed only with your permission or as required by law. Confidentiality will be maintained by means of ascribing a coded number to each participant when analysing the data. Only the researcher and her supervisor will have access to the collected data. The transcribed data will be kept safe in the supervisor's office in a locked drawer until analyses of the information has been completed and will then be destroyed and discarded appropriately.

**What will happen in the unlikely event of some form injury occurring as a direct result of your taking part in this research study?**

Not applicable

**Will you be paid to take part in this study and are there any costs involved?**

You will not be paid to take part in this study. There will be no costs involved for you, if you do take part.

**Is there anything else that you should know or do?**

- You can contact Dr Chrisma Pretorius at tel 021-8083453 if you have any further queries or encounter any problems.
- You can contact the Health Research Ethics Committee at 021-938 9207 if you have any concerns or complaints that have not been adequately addressed by your study doctor.
- You will receive a copy of this information and consent form for your own records.

### Declaration by participant

By signing below, I ..... agree to take part in a research study entitled (The lived experience of individuals living with Huntington's disease in the Western Cape, South Africa).

I declare that:

- I have read or had read to me this information and consent form and it is written in a language with which I am fluent and comfortable.
- I have had a chance to ask questions and all my questions have been adequately answered.
- I understand that taking part in this study is **voluntary** and I have not been pressurised to take part.
- I may choose to leave the study at any time and will not be penalised or prejudiced in any way.
- I may be asked to leave the study before it has finished, if the study doctor or researcher feels it is in my best interests, or if I do not follow the study plan, as agreed to.

Signed at (*place*) ..... on (*date*) ..... 2013.

.....  
**Signature of participant**

.....  
**Signature of witness**

### Declaration by investigator

I (*name*) ..... declare that:

- I explained the information in this document to .....

- I encouraged him/her to ask questions and took adequate time to answer them.
- I am satisfied that he/she adequately understands all aspects of the research, as discussed above
- I did/did not use a interpreter. *(If an interpreter is used then the interpreter must sign the declaration below.*

Signed at (*place*) ..... on (*date*) ..... 2013.

.....  
**Signature of investigator**

.....  
**Signature of witness**

#### **Declaration by interpreter**

I (*name*) ..... declare that:

- I assisted the investigator (*name*) ..... to explain the information in this document to (*name of participant*) ..... using the language medium of Afrikaans/Xhosa.
- We encouraged him/her to ask questions and took adequate time to answer them.
- I conveyed a factually correct version of what was related to me.
- I am satisfied that the participant fully understands the content of this informed consent document and has had all his/her question satisfactorily answered.

Signed at (*place*) ..... on (*date*) .....(2013)

.....  
**Signature of interpreter**

.....  
**Signature of witness**

### Appendix 3

#### DEELNEMERINLIGTINGSBLAD EN -TOESTEMMINGSVORM

**TITEL VAN DIE NAVORSINGSPROJEK: Die beleefde ervarings van individue met Huntington se siekte in die Wes-Kaap, Suid-Afrika**

**VERWYSINGSNOMMER: S13/04/087**

**HOOFNAVORSER: Ninon Joubert**

**ADRES: 9 Brenton Crescent, Belvedere Estate, 7550**

**KONTAKNOMMER: 0725456678**

U word genooi om deel te neem aan 'n navorsingsprojek. Lees asseblief hierdie inligtingsblad op u tyd deur aangesien die detail van die navorsingsprojek daarin verduidelik word. Indien daar enige deel van die navorsingsprojek is wat u nie ten volle verstaan nie, is u welkom om die navorsingspersoneel of dokter daaroor uit te vra. Dit is baie belangrik dat u ten volle moet verstaan wat die navorsingsprojek behels en hoe u daarby betrokke kan wees. U deelname is ook **volkome vrywillig** en dit staan u vry om deelname te weier. U sal op geen wyse hoegenaamd negatief beïnvloed word indien u sou weier om deel te neem nie. U mag ook te eniger tyd aan die navorsingsprojek onttrek, selfs al het u ingestem om deel te neem.

**Hierdie navorsingsprojek is deur die Gesondheidsnavorsingsetiekkomitee (GNEK) van die Universiteit Stellenbosch goedgekeur en sal uitgevoer word volgens die etiese riglyne en beginsels van die Internasionale Verklaring van Helsinki en die Etiese Riglyne vir Navorsing van die Mediese Navorsingsraad (MNR).**

#### **Wat behels hierdie navorsingsprojek?**

- Die doel van hierdie studie is om die beleefde ervarings van individue met Huntington se siekte (HS) in Suid-Afrika te verken. Die uitdagings sowel as die hulpbronne wat help om die siekte te hanteer sal ondersoek word.
- Dit is belangrik om hierdie navorsing te doen omdat HS 'n aftakelende lewenslange siekte is, wat fisiese en sielkundige simptome inhou vir die individue met HS. 'n Paar studies het al die lewende ervaring en hulpbronne van individue met HS ondersoek, maar geen studies van hierdie aard is al in Suid-Afrika uitgevoer nie. Kennis van die

aard kan help met die ontwerp en implementering van intervensies vir individue met HS.

- Die navorsing sal plaasvind by jou huis of by 'n plek wat jou pas. Ten minste 15 individue sal ingesluit word in hierdie studie. Die data-insameling sal begin met die voltooiing van 'n biografiese vraelys. Daarna sal 'n semi-gestruktureerde onderhoud volg, wat sal bestaan uit vrae oor jou lewende ervaring met HS. Hierdie onderhoud sal tussen 60 en 90 minute duur. Indien jy instem sal die onderhoud op band opgeneem word, waarna dit verbatim getranskribeer sal word om te help met die proses van data-analise.

### **Waarom is u genooi om deel te neem?**

- Jy is uitgenooi om deel te neem aan hierdie studie omdat jy as 'n individue met HS geïdentifiseer is. Jy het ook belangstelling om deel te neem getoon by 'n HS-ondersteuning groepsbyeenkoms wat in Claremont op 7 Februarie 2013 gehou is.

### **Wat sal u verantwoordelikhede wees?**

- Jou verantwoordelikhede vir deelname in hierdie studie behels dat jy deelneem aan 'n eenmalige semi-gestruktureerde onderhoud, wat ongeveer 60 tot 90 minute sal duur, om oor jou ervarings om met HS te lewe te praat. 'n Eenmalige fokusgroep, wat die ander deelnemers van hierdie studie sal insluit, om die resultate te verifieer, sal na die voltooiing van al die onderhoude geskeduleer word. Jou deelname aan die fokusgroep sal 'n waardevolle bydrae tot die studie maak. Indien jy sou wil deelneem aan die fokusgroep, sal die navorser u in kennis stel van die datum en plek.

### **Sal u voordeel trek deur deel te neem aan hierdie navorsingsprojek?**

- Jy sal nie direk voordeel trek deur deel te neem in hierdie studie nie. Die bevindinge van hierdie studie kan egter moontlik gepubliseer word in 'n wetenskaplike artikel in 'n portuurbeoordeelde joernaal. Die publikasie van die bevindinge van hierdie studie sal hopelik die eerste stap wees tot die bewusmaking van die ervaring van individue met Huntington se siekte in 'n Suid-Afrikaanse konteks en ook toekomstige studies stimuleer op hierdie gebied.

### **Is daar enige risiko's verbonde aan u deelname aan hierdie navorsingsprojek?**

- Die enigste voorsienbare risiko wat jy as deelnemer mag ervaar, is emosionele omgemak tydens die onderhoud as gevolg van die persoonlike aard van die onderhoud. Sou dit die geval wees, sal ek die deelnemers verwys na my studieleier, Dr. Chrisma Pretorius, n Voorligtingsielkundige, wie jou sal verwys na 'n toepaslike professionele gesondheidsorg werker. Haar kontakbesonderhede is: 021-808 3453 of [chrismapretorius@sun.ac.za](mailto:chrismapretorius@sun.ac.za).

**Watter alternatiewe is daar indien u nie instem om deel te neem nie?**

- Hierdie studie sluit nie enige vorm van behandeling in nie, dus sal geen alternatief aangebied word nie.

**Wie sal toegang hê tot u mediese rekords?**

- Enige inligting wat deur middel van hierdie studie verkry word en wat geïdentifiseer kan word met jou sal vertroulik bly en sal slegs met jou toestemming of soos wat deur die wet vereis word, openbaar word. Vertroulikheid sal gehandhaaf word deur middel van 'n nommer toe te ken aan elke deelnemer by die data ontleding. Slegs die navorser en haar studieleier sal toegang hê tot die data. Die getranskriebeerde data sal veilig bewaar word in die studieleier se kantoor in 'n toegesluite laai totdat die ontleding van die inligting afgehandel is en sal dan in 'n gepaste gewyse vernietig word.

**Wat sal gebeur in die onwaarskynlike geval van 'n besering wat mag voorkom as gevolg van u deelname aan hierdie navorsingsprojek?**

- Nie van toepassing nie.

**Sal u betaal word vir deelname aan die navorsingsprojek en is daar enige koste verbonde aan deelname?**

U sal nie betaal word vir deelname aan die navorsingsprojek nie. Deelname aan die navorsingsprojek sal u niks kos nie.

**Is daar enigiets anders wat u moet weet of doen?**

- U kan dr Chrisma Pretorius kontak by tel 021-8083453 indien u enige verdere vrae het of enige probleme ondervind.
- U kan die **Gesondheidsnavorsingsetiek administrasie** kontak by 021-938 9207 indien u enige bekommernis of klage het wat nie bevredigend deur u studiedokter hanteer is nie.
- U sal 'n afskrif van hierdie inligtings- en toestemmingsvorm ontvang vir u eie rekords.

## Verklaring deur deelnemer

Met die ondertekening van hierdie dokument onderneem ek,  
 ..... , om deel te neem aan 'n navorsingsprojek  
 getiteld (**Die beleefde ervarings van individue met Huntington se siekte in die Wes-  
 Kaap, Suid-Afrika**).

### Ek verklaar dat:

- Ek hierdie inligtings- en toestemmingsvorm gelees het of aan my laat voorlees het en dat dit in 'n taal geskryf is waarin ek vaardig en gemaklik mee is.
- Ek geleentheid gehad het om vrae te stel en dat al my vrae bevredigend beantwoord is.
- Ek verstaan dat deelname aan hierdie navorsingsprojek **vrywillig** is en dat daar geen druk op my geplaas is om deel te neem nie.
- Ek te eniger tyd aan die navorsingsprojek mag onttrek en dat ek nie op enige wyse daardeur benadeel sal word nie.
- Ek gevra mag word om van die navorsingsprojek te onttrek voordat dit afgehandel is indien die studiedokter of navorser van oordeel is dat dit in my beste belang is, of indien ek nie die ooreengekome navorsingsplan volg nie.

Geteken te (*plek*) ..... op (*datum*) ..... 2013.

.....  
**Handtekening van deelnemer**

.....  
**Handtekening van getuie**

## Verklaring deur navorser

Ek (*naam*) ..... verklaar dat:

- Ek die inligting in hierdie dokument verduidelik het aan  
 .....
- Ek hom/haar aangemoedig het om vrae te vra en voldoende tyd gebruik het om dit te beantwoord.

- Ek tevrede is dat hy/sy al die aspekte van die navorsingsprojek soos hierbo bespreek, voldoende verstaan.
- Ek 'n tolk gebruik het/nie 'n tolk gebruik het nie. (*Indien 'n tolk gebruik is, moet die tolk die onderstaande verklaring teken.*)

Geteken te (*plek*) ..... op (*datum*) ..... 2013.

.....  
**Handtekening van navorder**

.....  
**Handtekening van getuie**

### **Verklaring deur tolk**

Ek (*naam*) ..... verklaar dat:

- Ek die navorser (*naam*) ..... bygestaan  
het om die inligting in hierdie dokument in Afrikaans/Xhosa aan (*naam van deelnemer*) ..... te verduidelik.
- Ons hom/haar aangemoedig het om vrae te vra en voldoende tyd gebruik het om dit te beantwoord.
- Ek 'n feitelik korrekte weergawe oorgedra het van wat aan my vertel is.
- Ek tevrede is dat die deelnemer die inhoud van hierdie dokument ten volle verstaan en dat al sy/haar vrae bevredigend beantwoord is.

Geteken te (*plek*) ..... op (*datum*) ..... 2013.

.....  
**Handtekening van tolk**

.....  
**Handtekening van getuie**



## Appendix 4

### Biographical information

Instructions: Please complete the biographical form. Circle the correct answer where applicable.

Name and Surname:.....

Age:.....

Gender: Male/Female

Ethnicity: Black / Coloured/ Indian/ White / Other\*

If other, please specify:.....

Home language: .....

How long ago were you diagnosed with Huntington's disease? .....

Do you belong to a support group? Yes/ No

If yes, for how long?.....

## Appendix 5

### Biografiese inligting

Instrukties: Voltooi asb u biografiese inligting. Dui u keuse aan deur die regte opsie te omring.

Naam en Van:.....

Ouderdom:.....

Geslag: Manlik/Vroulik

Etnisiteit: Blank / Kleurling/ Indiër/ Swart / Ander\*

Indien ander, spesifiseer asb:.....

Huistaal: .....

Hoe lank gelede is u met Huntington se siekte gediagnoseer? :.....

Is u deel van 'n ondersteuningsgroep? Ja / Nee

Indien u is, vir hoe lank?.....

## **Appendix 6**

English: Questions for participants

1. Tell me about yourself and your family.
2. Tell me about the process of getting to a diagnosis of HD
3. Tell me about your experience of living with HD
4. What are some of the challenges that you face? Or What makes it difficult for you to cope with HD (What about Huntington's disease makes life harder? What symptoms of HD have you experienced so far?)
5. What are the factors and/resources that make it easier for you to cope with HD?
6. How does the progression of HD relate to the challenges and supports that you experience?
7. Do you have any other family members with HD? What has it been like having to deal with their HD also?
8. How do you see your future living with HD?
9. What was life like for you before HD? How would you describe it?

## **Appendix 7**

Afrikaans: Vrae vir deelnemers

1. Vertel my oor jouself en jou familie.
2. Vertel my oor die proses van die diagnose van Huntington se siekte (HS).
3. Vertel my oor jou ervaring om te lewe met HS.
4. Wat is die struikelblokke wat jy ervaar? Of wat maak dit moeilik vir jou om HS te hanteer?
5. Watter faktore en/of hulpbronne maak dit makliker vir jou om HS te hanteer.
6. Hoe hou die verloop van HS verband met die uitdagings en ondersteuning wat jy ervaar?
7. Het jy enige ander familie-lede met HS? Vertel my hoe dit voel dat jy hulle HS ook moet hanteer.
8. Hoe sien jy jou toekoms?
9. Hoe was lewe voor HS? Hoe sal jy dit beskryf?

## Appendix 8



UNIVERSITEIT•STELLENBOSCH-UNIVERSITY  
jou kennisvennoot • your knowledge partner

### Approval Notice Response to Modifications- (New Application)

06-Jun-2013  
JOUBERT, Ninon

Ethics Reference #: S13/04/087

Title: The lived experience of individuals living with huntingtons disease in the western cape , south Africa

Dear Miss Ninon JOUBERT,

The Response to Modifications - (*New Application*) received on 23-May-2013, was reviewed by members of Health Research Ethics Committee 2 via Expedited review procedures on 05-Jun-2013 and was approved.

Please note the following information about your approved research protocol:

Protocol Approval Period: 05-Jun-2013 -05-Jun-2014

Please remember to use your protocol number (S13/04/087) on any documents or correspondence with the HREC concerning your research protocol.

Please note that the HREC has the prerogative and authority to ask further questions, seek additional information, require further modifications, or monitor the conduct of your research and the consent process.

#### After Ethical Review:

Please note a template of the progress report is obtainable on [www.sun.ac.za/ethics](http://www.sun.ac.za/ethics) and should be submitted to the Committee before the year has expired.

The Committee will then consider the continuation of the project for a further year (if necessary). Annually a number of projects may be selected randomly for an external audit.

Translation of the consent document to the language applicable to the study participants should be submitted.

Federal Wide Assurance Number: 00001372

Institutional Review Board (IRB) Number: IRB0005239

The Health Research Ethics Committee complies with the SA National Health Act No.61 2003 as it pertains to health research and the United States Code of Federal Regulations Title 45 Part 46. This committee abides by the ethical norms and principles for research, established by the Declaration of Helsinki, the South African Medical Research Council Guidelines as well as the Guidelines for Ethical Research: Principles Structures and Processes 2004 (Department of Health).

#### Provincial and City of Cape Town Approval

Please note that for research at a primary or secondary healthcare facility permission must still be obtained from the relevant authorities (Western Cape Department of Health and/or City Health) to conduct the research as stated in the protocol. Contact persons are Ms Claudette Abrahams at Western Cape Department of Health ([healthres@pgwe.gov.za](mailto:healthres@pgwe.gov.za) Tel: +27 21 483 9907) and Dr Helene Visser at City Health ([Helene.Visser@capetown.gov.za](mailto:Helene.Visser@capetown.gov.za) Tel: +27 21 400 3981). Research that will be conducted at any tertiary academic institution requires approval from the relevant hospital manager. Ethics approval is required BEFORE approval can be obtained from these health authorities.

We wish you the best as you conduct your research.

For standard HREC forms and documents please visit: [www.sun.ac.za/ethics](http://www.sun.ac.za/ethics)

If you have any questions or need further assistance, please contact the HREC office at 0219389207.

#### Included Documents:

IC FORM  
BIOGRAFIESE INLIGTING  
LETTER  
IC FORM  
QUESTIONS  
DEMOGRAPHIC  
COV LETTER  
DEC LETTER JOUBERT  
SYNOPSIS  
CHECKLIST

PROTOCOL  
CV JOUBERT  
BUDGET  
DEC LETTER PRETORIUS  
APPLIC FORM  
VRAE  
CV PRETORIUS

Sincerely,

Mertrude Davids  
HREC Coordinator  
Health Research Ethics Committee 2